Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship forzen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
15 Mold Panel	1465			In-Common Laboratories	Done at Alletess			12-Mar-20
2,3-Dinor-11Beta-Prostaglandin F2 Alpha, 24 Hour, Urine [23BPT]	1		Urine. No preservative. Collect for 24-h or random sample. Refrigeate 4°C during collection. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	systemic mastocytosis; mast cell activation disorders including systemic mastocytosis; Replaces 11 BETA-PROSTAGLANDIN F2 ALPHA	LC-MS/MS	Replaced by Mast Cell Mediators. May also order Leukotriene E4	23-Jul-24
21-Hydroxylase Antibodies, Serum [21OHAB]			Red Top. Store serum frozen	In-Common Laboratories	autoimmume adrenal failure	Available at CHUM	Semt to USA	
21-Hydroxylase Antibodies, Serum [OH21]			Red Top. Store serum frozen	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	autoimmume adrenal failure	Available at CHUM		
34-plex Cytokine Panel	1523		Red To tube only. (Serum.) Store at -20°C.	OncoHelix			FIRST CHOICE	26-Nov-24
5-Methyltetrahydrofolate (CSF) [NC01]			Collect 1 milliliter of CSF. Spin sample if contaminated with red blood cells and freeze the clear CSF at -80°Celsius. Store frozen at -80°Celsius.	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Cerebral folate deficiency			14-May-20
5'Nucleotidase	4		Serum. Stable 4h at RT, 7 days at 4°C or 14 days at -20°C.	Dynacare		enzymatic	Requires preappova as tets is of limited utility	22-Oct-20
68KD (HSP-70) Antibodies [F68KD]	1280		Red Top (preferred); Gold SST. Store at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	autoimmune hearing loss	No longer available at CHUQ: Anticorps anti-oreille interne (anticochlee)		3-May-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
8-hydrox-acyclovir and 9-carboxymethy guanosine	1465		Serum. Red Top preferred over gel.	Karolinska University Hospital at Huddinge Clinical Pharmacology Laboratory, C2:69 SE-141 86 Stockholm		LC-MS/MS		8-Jan-22
A-ß (beta) 42/40	140		For CSF, use Sarstedt CSF Tube 62.610.018. Collect CSF directly into Sarstedt tubes and fill the tube 50% to 80% minimum. The specimen must not be aliquoted from a regular collection tube. If the first 1mL collected is hemolyzed, discard and continue collection with a new tube.	BC Neuroimmunology (bcneuro.ca)	Less discimination than p217 on SIMOA		Requires: Neurodegenerative Profile Requisition Form	6-Jun-24
a1-Acid glycoprotein [A1AGP]			Serum. Freeze if > 72 hours	In-Common Laboratories	Other names: Orosomucoid, Acid Glycoprotein, Alpha 1 Acid Glycoprotein	Nephelometry		6-Sep-18
Acetaminophen [ACETA]			Serum	In-Common Laboratories	quantitation; to determine clearance			6-Sep-18
Acetylcholine Receptor Antibodies (AChR Ab, 91020/91021)	18		Serum (Gold SST) ONLY. Store at -20°C.	BC Neuroimmunology (bcneuro.ca)	myasthenia gravis	RIPA -first performed 91020 qualitative test. If positive tham 91021 quantitative test		27-Mar-25
Acetylcholine Receptor Antibodies with reflex to Muscle Specific Tyrosine Kinase Antibodies (MuSK Ab)	18		Serum (Gold SST) ONLY. Store at -20°C.	BC Neuroimmunology (bcneuro.ca)	myasthenia gravis	RIPA		9-Aug-22
Acetylcholine Receptor Antibody [ACRAB]	18		Red Top; Gold SST. Store and send serum at -20°C.	In-Common Laboratories	myasthenia gravis	RIA	CHUS: Ac anti Récepteur acétylcholine	17-Feb-23
ACUTE PROMYELOCYTIC LEUKEMIA, INDUCER OF; PML (102578)		PML (102578)	Lavender Top (EDTA)	Jewish General	Leukemia, acute promyelocytic, PML/RARA type		PML/RARA FUSION GENE, INCLUDED	

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Acyclovir, Serum/Plasma [0158SP]	1325		Red Top or Lavender Top (NOT SST). Store frozen at -20°C.	NMS Laboratories		LC-MS/MS		27-Apr-22
Adalimumab	10		Serum. Stable 48 h at RT, 5 days at 4°C, or 30 days at -20°C.	Dynacare Next	Humira®	ELISA	available at HMR	24-Oct-18
Adalimumab (Anser ADA) [3170]	10		Gold SST or Red Top	Prometheus Therapeutics and Diagnostics		Quote 98765	Dynacare Next; availableat HMR	24-Oct-18
ADAR Full Gene Sequencing Analysis [MOL309]		ADAR (146920)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Aicardi-goutieres syndrome 6 (615010); Dyschromatosis symmetrica hereditaria	Sanger sequencing	NGS Panel available	14-May-20
Adenosine Deaminase and Purine Nucleoside Phosphorylase	11		Guthere card + 1 mL EDTA plasma. Keep at RT.	Michael Hershfield, M.D. Room 235 Sands Building 303 Research Dr, Duke Univ Med Cent Durham, North Carolina 27710 USA Tel 1: 919-684-4184 Tel 2: 919-684-2622				12-Nov-25
Adenosine Deaminase, Pleural Fluid [FADPF]	11		Must be frozen within 24 hours of collection. No freeze/thaw cycles.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Ultraviolet Spectrophotometry		11-Feb-21
Adiponectin [FADIO]			Draw blood in a plain red-top or Gold SST, serum gel tube(s) is acceptable. Spin down and send 1 mL of serum refrigerated in a plastic vial. Store frozen. Overnight fasting is required.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELSIA	HIC: RUO	31-Oct-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Admark® Alzheimer's Evaluation, CSF (FDA Cleared) [92433]{Replaces: ADmark® Phospho-Tau/Total-Tau/Ab42 Analysis & Interpretation, CSF (Symptomatic) [177]}	596	Beta-Amyloid (1-42), CSF tTau, CSF pTau181, CSF tTau/Abeta42 Ratio pTau181/Abeta42 Ratio	Cerebrospinal Fluid (CSF), 1 mL. Only acceptable tube: CSF tube 63.614.625 (Sarstedt). Specimen Stability Room temperature: 5 days Refrigerated: 14 days Frozen: 56 days.	Quest Diagnostics	Alzheimer Disease (104300)	ECLIA	Has special draw instructions.	22-Dec-25
Adrenal hyperplasia due to 21-hydroyxlase deficiency (201910)	1326	CYP21A2 (613815)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		Sanger sequencing & MLPA	SEND to BLUEPRINT for CAH Panel	23-Jan-25
Afirma GEC	1680		Fine needle aspirate	Groupe TMTC (https://grouptmtc.com)			ON HOLD. Requires approval by head of OOP	21-Jan-21

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Agammaglobulinemia Panel, Sequencing (9 Genes) and Deletion/Duplication (6 Genes) [2011151]	999	BLNK, BTK, CD79A, CD79B, IGHM, IGLL1, LRRC8A, PIK3R1, SH2D1A	Lavender Top (EDTA)	ARUP Laboratories (www.aruplab.com)	Agammaglobulinemia 1, Autosomal Recessive ◦ X-Linked Agammaglobulinemia ◦ SH2D1A-Related Lymphoproliferative Disease, X-Linked ◦ Agammaglobulinemia 2, Autosomal Recessive ◦ Agammaglobulinemia 3, Autosomal Recessive ◦ Agammaglobulinemia 4, Autosomal Recessive ◦ Agammaglobulinemia 5, Autosomal Dominant ◦ Agammaglobulinemia 6, Autosomal Recessive ◦ Agammaglobulinemia 7, Autosomal	Massively Parallel Sequencing/Exonic Oligonucleotide-based CGH Microarray		
AGXT Gene, Full Gene Analysis [AGXMS]		AGXT (604285)	Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Hyperoxaluria, primary, type 1 (259900)	Sanger sequencing and MLPA		

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
aHUS (complement mediated TMA) Panel [aHUS-FP]	1306	C3, C3c, C4, C5, FD, FB, Ba, Bb, properdin, soluble C5b-9, Hi, FH, CH50, APFA, FHAA, FBAA, C3b deposition, Fluid pahse activity-IFE	2 mL serum RED TOP ONLY + 2 mL frozen EDTA plasma. Store frozen.	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)			replaces TMA Funcional Panel	1-Mar-25
Aicardi-Goutieres Syndrome (NextGen Sequencing Panel and Copy Number Analysis; 6 Genes) [NGS344]		ADAR (601059); ALDH7A1 (107323); RNASEH2A (606034); RNASEH2B (610326); RNASEH2C (610330); SAMHD1 (606754); TREX1 (606609)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Aicardi-Goutieres Syndrome	NextGen Sequencing		14-May-20
ALA Dehydratase [ALAD]			Green Top (Na heparin), 4°C only.Do not freeze. Send M-W only.	In-Common Laboratories	Aminolevulinic Acid Dehydratase Deficiency Porphyria (612740)		Sent to USA	6-Sep-18
Albendazole	1917		RED TOP ONLY. Serum without gel, centrifuge immediately after collection, freeze and ship with dry ice	Inselspital Bern Freiburgstrasse 10 Zentrum für Labormedizin Zentrale Annahme INOF / z. Hd. Y. Aebi CH-3010 Bern, Switzerland	Measures albendazole sulfoxide due to 1st pass effcets			28-Feb-25
Albright's hereditary Osteodystrophy (103580)	440	GNAS (139320)	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)	Albright's hereditary Osteodystrophy (103580)	NextGen Sequencing		26-Mar-25
Alexander Disease via the GFAP Gene [3775]	979	GFAP (137780)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Alexander disease (203450)	Sanger sequencing		10-Jul-23
Alpha aminoadipic semialdehyde (Urine) [MET20]	12	Alpha-aminoadipic semialdehyde	Collect 1 milliliter urine (random) and freeze at -20°Celsius. Store frozen at –20°Celsius and ship frozen.	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Pyridoxine-dependent epilepsy (266100)	MS/MS		14-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Alpha aminoadipic semialdehyde (Whole Blood) [AASA]	12	Alpha-aminoadipic semialdehyde	Specimen: Whole Blood Container(s): Dark Green/Sodium Heparin or Lt. Green/Lithium Heparin Tube. **Reject due to**: If sample is not spun and frozen within 1 hour of collection. 48 hr storage at -20°C is acceptable. Store **plasma** at -70°C for 6 months.	Seattle Children's Hospital. http://seattlechildrenslab.testcatalog.org/	Pyridoxine-dependent epilepsy (266100)			29-May-25
Alpha Thalassemia Sequence HBA1 and HBA2 330 (Unknown alpha-chain Hemoglobin Variants)	820	HBA1 (141800)/HBA2 (141850)	Lavender Top (EDTA)	Clinical Genetics Laboratory, Juravinski Hospital, Hamilton, ONT	also availble at CHUM		Review AH612 for test sought as req is non-specific	10-Nov-25
Alpha-1-Antitrypsin Clearance, Feces and Serum [A1AFS]	1465		Both feces and serum are required. Blood must be drawn during the stool collection period.**Specimen Type**: Serum Collection Container/Tube: Red top or serum gel. Collection Instructions: Centrifuge within 2 hours. **Specimen Type: Feces** Supplies: Stool Containers - 24, 48, 72 Hour Kit (T291) Container/Tube: Stool container Specimen Volume: Entire collection Collection Instructions: 1. Collect a 24-hour fecal collection. 2. If no specimen is obtained within 24 hours, extend collection time to 48 to 72 hours. Stable for 14 d at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	protein-losing enteropathy	Immunonephelometry		25-Oct-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
ALPHA-1-ANTITRYPSIN DEFICIENCY (613490)	1395	SERPINA1 (104400)	Lavender Top (EDTA)	Attn: Norine Freedman or Lynn Coleman Special Chemistry Laboratory St Paul's Hospital 1081 Burrard Street Vancouver, B.C. V6Z 1Y6. Contact Dr. A. Mattman BEFORE sending sample.	ALPHA-1-ANTITRYPSIN DEFICIENCY (613490)	Sanger sequencing	Send to Fulgent.	
ALPHA-1-ANTITRYPSIN DEFICIENCY (SERPINA1 Single Gene Test) (613490)	1395	SERPINA1 (104400)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)	ALPHA-1-ANTITRYPSIN DEFICIENCY (613490)		Preferred	26-Mar-25
Alpha-1-Antitrypsin, Random, Feces [A1AF]	1465		Homogenized Stool: 1 mL. Stable for 14 d at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	protein-losing enteropathy	Immunonephelometry	Surrogate Marker	25-Oct-24
Alpha-1-Antttrypsin, Random, Feces [A1AF]	1465		Random fecal sample (5 g). Store frozen for up to 14 days	Mayo Clinical Laboratories (https://www.mayocliniclabs.com/index.html)	Immunonephelometry			20-Apr-23
Alpha-aminoadipic semialdehyde (CSF) [NC08]	12	Alpha-aminoadipic semialdehyde	Collect 1 milliliter of CSF. Spin sample if contaminated with red blood cells and freeze the clear CSF at -80°Celsius. Store frozen at -80°Celsius.	MNG Laboratories (www.medicalneurogenetics.com)/Dyncare	Pyridoxine-dependent epilepsy (266100)	MS/MS	Seattle for Heparin plasma	14-May-20
Alpha-Subunit Pituitary Tumor Marker, Serum [APGH]	55		Red Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Alternate name: Alpha Glycoprotein Subunit			19-Dec-24
Alpha-Thalassemia (604131)	826	HBA1 (141800); HBA2 (141850)	Lavender Top (EDTA)	Molecular Genetics Laboratory - McMaster University Medical Centre	targeted mutations at HSJ	Sanger sequencing & del/dupl	Also available at CHUM	23-Jan-23
Alport Syndrome NGS Panel	785	COL4A3, COL4A4, COL4A5 COL4A6	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		26-Mar-25
Alzheimer Disease, Familial or Cerebral Amyloid Angiopathy via the APP Gene [4557]	598	APP (104760)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NGS	For known mutations only. Otherwise use Fulgent panel.	10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
AMELOGENESIS IMPERFECTA VIA THE DLX3 GENE [9061]		DLX (600525)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Tricho-Dento-Osseous Syndrome (190320); Amelogenesis Imperfecta, Type IV (104510)	NextGen Sequencing		10-Jul-23
Aminolevulinic Acid Dehydratase (ALAD), Whole Blood [ALAD]			Green Top, 4°C only	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Aminolevulinic Acid Dehydratase Deficiency Porphyria (612740)			31-Oct-20
AMYLOID PROTEIN ID, PAR, LC MS/MS [AMPIP]	1123		Paraffin section	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS	ADD CHARGE FOR MICRODISSECTION $1216.10 [MLCPC] & MS $259.90 [MSPTC]	6-Jun-25
Amyloid Related Disorders [NGS380]	1377		Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NGS		21-Mar-23
Amyotrophic Lateral Sclerosis / Motor Neuron Disease via the FUS Gene [6927]	853		Lavender Top (EDTA) 2 x 4 mL	Prevention Genetics (www.preventiongenetics.com)		NextGen Sequencing	Transferred to MUHC	4-Oct-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Panel [10359]	853	ANG ANXA1APP ARHGEF28 ATXN2 C9orf72 CFAP410 CHCHD10 CHMP2B DAO DCTN1 ERBB4 FIG4 FUS GRN HNRNPA1 HNRNPA2B1 ITM2B KIF5A MAPT MATR3 MOBP NEFH 9 NEK1 OPTN PFN1 PSEN1 PSEN2 SETX SOD1 SQSTM1 TAF15 TARDBP TBK1 TREM2 TUBA4UBQLN2 UNC13A VAPB VCP	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Frontotemporal Dementia And/Or Amyotrophic Lateral Sclerosis (105550), Amyotrophic Lateral Sclerosis Type 6 (608030); Amyotrophic Lateral Sclerosis Type 9 (611895); Amyotrophic Lateral Sclerosis Type 1 (105400); Amyotrophic Lateral Sclerosis Type 10 (612069); Amyotrophic Lateral Sclerosis Type 12 (613435)	NextGen Sequencing		10-Jul-23
Andersen-Tawil syndrome (170390)		KCNJ2 (600681)	Lavender Top (EDTA)	Invitae (www.invitae.com)	ANDERSEN CARDIODYSRHYTHMIC PERIODIC PARALYSIS (proper name); Andersen syndrome (170390), long QT syndrome 7, PERIODIC PARALYSIS, POTASSIUM-SENSITIVE CARDIODYSRHYTHMIC TYPE			10-Jul-23
Anemia Panel [HE0401]	1582		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NGS		12-Nov-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Angelman Syndrome: Methylation and Copy Number Analysis	311	SNRPN	Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children		Methylation-Specific-MLPA of SNPRN		19-Feb-20
Angelman Syndrome: UPD 15 Analy	311		Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children		STR analysis		19-Feb-20
Angiotensin Converting Enzyme (ACE), CSF	1919		CSF. Store Frozen	In-Common Laboratories				
Anser IFX (infliximab) [3150]			Gold SST or Red Top	Prometheus Therapeutics and Diagnostics	Remicaide®	Quote 98765		
Anti-AAV9 Antibody Screening	1465		Serum. Use Gold SST. Store frozen.	Viroclinics Biosciences	Use NPC			12-Dec-22
Anti-Aquaporin 4 (Anti-AQP4 Ab) + Anti-Myelin Oligodendrocyte Glycoproteins antibody (Anti-MOG)	157		Serum ONLY (Red Top OR Gold SST). Store at -20°C. (CSF is not an appropriate sample as Ab is made by plasma cells peripheraly.)	BC Neuroimmunology (bcneuro.ca)	Anti-Aquaporin 4 (Anti-AQP4 Ab) $125; Anti-Myelin Oligodendrocyte Glycoproteins antibody (Anti-MOG) $125	CBA live. Confirmation by IHC.	NMO avaialble at CHUM for CSF and Serum as "NMO-Ig". Only on rare occasions, MOG testing is done on CSF as its utility is uncertain; serum is 1st choice.	18-Apr-24
Anti-AT1R Antibody	1465		Serum. Store frozen.	London Laboratories Service Group - Transplant Laboratory				31-Aug-21
Anti-C1Q Ab, IgG, Serum [C1QABS]	1402		Serum. Store at -20°C. Stable 1 year.	In-Common Laboratories		Semi-quantitative ELISA		25-Nov-25
Anti-DNAse B Titer, Serum [ADNAS]	1465		Serum (Gold or Red Top). Stable for 28 days at 4°C (preferred) or -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Nephelometry		23-Jul-23
Anti-Domain 1 Beta-2 Glycoprotein 1 (D1 ß2GP1) (IgG only)	1572	Serum		Mitogen Advanced Diagnostics	CIA			13-Jan-26
Anti-dsDNA (Double-stranded) Ab by Farr method (RDL) [520059]	1465		Serum (Red Top or Gold SST). Store at -20°C.	LabCorp/Dynacare (https://www.labcorp.com/)		RIA	Only if specifically requestedby this method	23-Nov-20
Anti-DSF70	2004			Mitogen				13-Jan-26

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Anti-**Elaprase** antibody	1406		Draw blood before infusion for patients on treatment. If a specimen is being drawn due to an infusion reaction, please wait between 4 and 24 h to allow ERT to clear from blood. Collect 6 ml of blood (3 ml serum to send frozen the day of collection ; if not possible to send it the same day of collection keep frozen at - 80C until shipping)	LabCorp (https://www.labcorp.com/)	Antibody produced in patients on Enzyme Replacement Therapy for Hunter syndrome (MPSII)			
Anti-Enterocyte Antibodies, Serum Analysis	1404		Red Top - Serum. Stable 1 week at 4°C. Store frozen.	The Children's Hospital of Philadelphia (https://www.testmenu.com/chop)				28-Jan-25
Anti-GAD 65 antibodies	29		CSF, Gold SST	Mitogen Advanced Diagnostics	stiff man syndrome	CBA	ANTICORPS ANTI-GAD65 (anti-glutamic acid decarboxylase 65kd) @ CHUM. SERUM and CSF.	21-Sep-23
Anti-GMCSF Autoantibodies [GMCSFA]	1279		Serum gel (Gold top); Freeze at -70°C.	National Jewish Health Laboratories		ELISA		23-May-19
Anti-IFNg Autoantibody [IFNGE]	1147		Gold SST or Red Top. 2-8°C for 48 hours, >1 month at -70°C	National Jewish Health Laboratories				18-Mar-21
Anti-IgA [FIGA]	908		Gold SST	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				4-Mar-21
Anti-IgLON5	1465		Gold SST	BC Neuroimmunology (bcneuro.ca)		CBA		18-Apr-24
anti-NMDA [Anti-récepteur glutamate NMDA (N-méthyl-D-aspartate)]			CSF only (Ab is made by plasma cells in CSF). Serum test available.	HSJ		IF	HSJ	20-Apr-18
Anti-Nucleolar Envelope/Membrane Test Panel	1741			Mitogen			RUO	13-Jan-26

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Anti-Phospholipase A2 Receptor (anti-PLA2R)	49		Gold SST	Mitogen Advanced Diagnostics	Membranous Nephropathy/Nephritis; MEMBRANOUS NEPHROPATHY, SUSCEPTIBILITY TO; MBNP (%614692)	CBA	Avaialble at HMR	21-Sep-23
Anti-phosphotidylserine/prothrombin complex (PS/PT) (IgG+IgM)	1572	Serum		Mitogen Advanced Diagnostics	ELISA			13-Jan-26
Anti-TB Drug	58	Azithromycin; Ethambutol; Rifabutin; Isonazid; Pyrazinamide; Rifampin	Serum	National Jewish Health Laboratories	TDM	LC/GCMS/LCMS	Same price for all anti-TB drugs per determination	23-Feb-24
Anti-THSD7A (thrombospodin)	1465		Gold SST; store serum at -20°C	Mitogen Advanced Diagnostics	primary membranous	CBA		13-Jan-26
Antibodies to clustered acetylcholine receptors (AChR Ab CBA)	1465		Serum (Gold SST). Store at -20°C.	BC Neuroimmunology (bcneuro.ca)	myasthenia gravis	CBA		31-Oct-20
Anticorps anti-21 hydroxylase			Gold SST; store serum at -20°C	CHUM	autoimmume adrenal failure			
Apixiban (anti-X)			Light Blue (2 tubes)	Biochemistry - JGH				7-Aug-18
Aripiprazole (Abilify) [FARI]	1142		Red top tube (Gold top SST tube not accepted)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		5-May-21
Arrhythmia Panel [CA1601]	289	ABCC9 AKAP9 ANK2 CACNA1C* CACNB2 CALM1* CALM2 CALM3 CASQ2 CAV3 CDH2 CTNNA3 DBH DSC2 DSG2 DSP C FLNC* GATA6 HADHA HCN4 KCNA5 KCNE1 KCNH2 KCNJ2 KCNJ5 KCNQ1 LDB3 LEMD2 LMNA MYH6 MYH7 MYL4 NKX2-5 NOS1AP PKP2#* PLN PPA2 RYR2 SALL4 SCN10A SCN1B SCN3B SCN5A TBX5 TECRL TGFB3 TMEM43 TNNI3 TNNI3K TNNT2 TRDN TRPM4 TTN*	Lavender Top (EDTA) [2 tubes]	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		20-Feb-20
Arthritis Panel	1465		Serum gel (Gold top)	Mitogen Advanced Diagnostics				13-Jan-26

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Aspergillus Antibody by Immunodiffusion [324]			Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial.STABILITY Refrigerated: 14 days Frozen: 6 months	MiraVista Diagnostics			Performed at CHUM. For Microbiology.	8-Sep-22
Ataxia Repeat Expansion Panel [4101]	583		Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories				8-Nov-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Ataxia/Episodic Ataxia Disorders (NextGen Sequencing Panel and Copy Number Analysis + mtDNA + FRDA Repeat Expansion Analysis) [NGS419]	583	AAAS (605378), AARS2 (612035), ABCB7 (300135), ABCC8 (600509), ABCD1 (300371), ABHD5 (604780), ACO2 (100850), ADCK3 (606980), ADSL (608222), AFG3L2 (604581), AHI1 (608894), ALDH5A1 (610045), ALG6 (604566), AMACR (NA), ANO10 (613728), APOPT1 (616003), APTX (606350), ARL13B (608922), ARL6 (608845), ARSA (607574), ASL (608310), ASS1 (603470), ATM (607585), ATN1 (607462), ATP1A2 (182340), ATP1A3 (182350), ATP8A2 (605870), ATPAF2 (608918), ATXN1 (601556), ATXN10 (603516), ATXN2 (601517), ATXN3 (607047), ATXN7 (607640), AUH (600529), B9D1 (614144), BBS1 (209901), BBS10 (610148), BBS12 (610683), BBS2 (606151), BBS4 (600374), BBS5 (603650), BBS7 (607590), BBS9 (607968), BCKDHA (608348), BCKDHB (248611), BCS1L (603647), BEAN1 (612051), BOLA3 (613183), BSCL2 (606158), BTD (609019), C10ORF2 (606075), C12orf65 (613541), C19orf12 (614298), C5ORF42 (614571), CA8 (114815), CACNA1A (601011), CACNA1G (604065), CACNB4 (601949), CAMTA1 (611501)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)		NextGen Sequencing		14-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Ataxia/Episodic Ataxia Disorders (NextGen Sequencing Panel and Copy Number Analysis + mtDNA) [NGS324]	583	AAAS (605378), AARS2 (612035), ABCB7 (300135), ABCC8 (600509), ABCD1 (300371), ABHD5 (604780), ACO2 (100850), ADCK3 (606980), ADSL (608222), AFG3L2 (604581), AHI1 (608894), ALDH5A1 (610045), ALG6 (604566), AMACR (NA), ANO10 (613728), APOPT1 (616003), APTX (606350), ARL13B (608922), ARL6 (608845), ARSA (607574), ASL (608310), ASS1 (603470), ATM (607585), ATN1 (607462), ATP1A2 (182340), ATP1A3 (182350), ATP8A2 (605870), ATPAF2 (608918), ATXN1 (601556), ATXN10 (603516), ATXN2 (601517), ATXN3 (607047), ATXN7 (607640), AUH (600529), B9D1 (614144), BBS1 (209901), BBS10 (610148), BBS12 (610683), BBS2 (606151), BBS4 (600374), BBS5 (603650), BBS7 (607590), BBS9 (607968), BCKDHA (608348), BCKDHB (248611), BCS1L (603647), BEAN1 (612051), BOLA3 (613183), BSCL2 (606158), BTD (609019), C10ORF2 (606075), C12orf65 (613541), C19orf12 (614298), C5ORF42 (614571), CA8 (114815), CACNA1A (601011), CACNA1G (604065), CACNB4 (601949), CAMTA1 (611501)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		14-May-20
Ataxia/Episodic Ataxia Disorders (NGS Panel and Copy Number Analysis + mtDNA+ SCA & FRDA Repeat Expansion Analysis) [NGS420]	583		Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing	No longer offered Alternate: NGS324 or MOL391	13-Sep-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Autism Spectrum Disorders and Intellectual Disability (ASD-ID) Comprehensive Sequencing Panel with CNV Detection [5045]	717	complete gene list available at https://www.preventiongenetics.com/documents/ASD_IDGeneList.pdf?07c00668	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Replaces Fulgent ID Panel when proband available only (adopted child…). If 'Trio' (proband + parents) send for Autism/ID Xpanded Panel at GeneDx (3500 USD for 3 samples)	NextGen Sequencing	Available at HSJ.	28-Nov-24
Autism/ID Xpanded Panel [952]	717		Lavender Top (EDTA)	GeneDx (www.genedx.com)		NextGen Sequencing	Available at HSJ.	4-Feb-25
Autoantibodies to interferon gamma [IFNGE]	1465		Serum (Red Top or Gold SST). Store at -70°C.	National Jewish Health Laboratories		ELISA		1-Oct-21
Autoimmune Dysautonomia Evaluation, Serum (ADE)			Red Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		IFA, RIA, EIA, WB, CBA		

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Autoimmune Liver Diseases Profile, Serum [ALDAB S]	1330		Gold SST	In-Common Laboratories	Anti AMA-M2 Ab (Anti Mitochondrial M2 antibody); Anti M2-3E (BPO) Ab; Anti Sp100 Ab(Anti Sp100 nuclear antigen antibody); Anti PML Ab (Anti Promyelocytic leukemia factor antibody); Anti gp210 Ab (Anti Glycoprotein 210 antibody); Anti LKM-1 (Cytochrome P450 II D6) Ab (Anti Liver kidney microsomal type 1 antibody); Anti LC-1 Ab (Anti Liver cytosol type 1 antibody); Anti SLA/LP Ab (Anti Soluble liver antigen/liver-pancreas antibody); Anti SSA (Ro60) Ab; Anti Ro52 (TRIM21) Ab; Anti Scl-70 Ab(Anti Topoisomerase I antibody); Anti	immunoblot	Mitogen Discontinued	30-Mar-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Autoimmune Myopathy /Myositis Profile PLUS (Includes Mup44 & Immune Mediated Necrotizing Profile)	153		Gold SST. Does not accept CSF.	Mitogen Advanced Diagnostics	Jo-1, Mi2, Mi2-α, Mi2β, MDA5, NXP2, TIF1γ PL7, PL12, PM/Scl75, PM/Scl100, Ku, SRP, EJ, OJ, Ro52, HMGCR, anti-NT5C1 A/Mup44	LIA, ELISA, ALBIA (LDT)		13-Jan-26
Autoimmune Polyendocrinopathy Syndrome Type 1 via the AIRE Gene [1224]		AIRE (607358)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Sanger sequencing		10-Jul-23
Autoimmune Retinopathy Panel by Immunoblot [ARP]	720		Serum. Store at 4°C. DO NOT FREEZE.	Ocular Immunology Laboratory, OHSU Biomedical Research Building, Room 253 3181 SW Sam Jackson Park Road Portland, OR 97239, USA	CAII (carbonic anhydrase II), HSP27 (heat shock protein 27), GAPDH (glyceraldehyde 3-phosphate dehydrogenase), Aldolase, Enolase, Arrestin, Tubulin, PKM2 (pyruvate kinase M2)	Immunoblot		17-Sep-24
Autoinflammatory and Autoimmunity Syndromes Panel [08120]	838		Lavender Top (EDTA)	Invitae (www.invitae.com)/Dynacare		NextGen Sequencing		28-Apr-25
Autoinflammatory Syndrome Panel [IM0201]	838		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		13-Jan-26
Avian IgG Antibodies Panel, Serum (Budgie and Pigeon)	1419		Red Top; Gold SST	In-Common Laboratories	budgie = parakeet	FEIA	Avian Precipitans not available in QC	10-Mar-20
Avian Precipitins: Budgie IgG Antibodies	1419		Red Top; Gold SST	In-Common Laboratories	Available at CHUQ (perruche/plumes); budgie = parakeet	FEIA		8-Nov-18
Avian Precipitins: Cockatiel	1419		Red Top; Gold SST	In-Common Laboratories	Available at CHUQ (perruche/plumes)	CIEP	ON HOLD	6-Sep-18
Avian precipitins: Parrot IgG Antibodies	1419		Red Top; Gold SST	In-Common Laboratories	Available at CHUQ (perroquet/plumes))	FEIA		8-Nov-18

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Avian precipitins: Pigeon IgG Antibodies	1419		Red Top; Gold SST	In-Common Laboratories		FEIA		8-Nov-18
Baller-Gerold syndrome (218600)		RECQL4 (603780)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Barium			Urine - metal free container	INSPQ	Alternate: Hospitals-In- Common			
Bartter Syndrome NGS Panel	804	Bartt	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)				26-Mar-25
Basal Ganglia Calcification NGS Panel	1192	CA2; MYORG; PDGFB (190040); PDGFRB; SLC20A2 (158378); XPR1	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)	Basal ganglia calcification, idiopathic, 5 (615483); Basal ganglia cancification, idiopathic, 1	NextGen Sequencing		3/26/2025
Beckwith-Wiedemann syndrome: CDKN1C Sequencing (Step 3)	571	CDKN1C (600856)	Lavender Top (EDTA) [2 tubes]	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Wiedemann-Beckwith Syndrome (130650)			19-Feb-20
Beckwith-Wiedemann syndrome: Methylation & Copy Number (Step1)	571		Lavender Top (EDTA) [2 tubes]	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Wiedemann-Beckwith Syndrome (130650)	Methylation-Specific-MLPA of imprinting centers 1 and 2		19-Feb-20
Beckwith-Wiedemann syndrome: UPD11 Analysis (Step 2)	571	H19 (103280); KCNQ1OT1 (604115); CDKN1C (600856)	Lavender Top (EDTA) [2 tubes]	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Wiedemann-Beckwith Syndrome (130650)	UPD11 studies via STR (short tandem repeat) analysis.		19-Feb-20
Beta Thalassemia Sequence HBB (Unknown beta-chain Hemoglobin Variants)	820	HBB (141900)	Lavender Top (EDTA)	Clinical Genetics Laboratory, Juravinski Hospital, Hamilton, ONT	also availble at MUHC		Review AH612 for test sought as req is non-specific	10-Nov-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Beta-2 Glycoprotein 1 Antibodies, IgM, Serum [MB2GP]	1572		Serum gel (Gold top)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Evaluation of suspected cases of antiphospholipid syndrome	ELISA	Available at CHUQ> IgA and IgM combined.. Stable for <h at RT. Store at -20°C.	1-Feb-23
Beta-Thalassemia (613985)	823	HBB (141900)	Lavender Top (EDTA)	Molecular Genetics Laboratory - McMaster University Medical Centre		Sanger sequencing	Also avaialble at MUHC (CMDL)	23-Jan-23
Bile Acids for Peroxisomal Disorders, Serum [BAIPD]	9		Serum. Store at 4°C or -20°C. Stable for 90 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		8-Nov-23
bipterin	763	CFTR (602421)	Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children (www.sickkids.ca/paediatriclabmedicinems/lab-divisions/genome-diagnostics/genome-diagnostics.html#genome)	Cystic Fibrosis (219700)	Sanger sequencing	Available at MUHC	8-Jan-20
Birt-Hogg-Dubé Syndrome (FLCN Single Gene Test)	740	FLCN (607273)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NGS		26-Mar-25
Blastomyces Antibody by EIA, Serum [BLAST]	491		Red Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Enzyme Immunoassay (EIA)	Reserved for Microbiology	31-Oct-20
Blastomyces Antibody by Immunodiffusion [322]	491		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. STABILITYRefrigerated: 14 days Frozen: 6 months	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Blastomyces Quantitative EIA [316]	491		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. Plasma: Collect plasma specimens in an EDTA, heparin or sodium citrate tube. Centrifuge for 15 minutes and pipette plasma into a plastic screw cap vial. Urine/CSF/BAL/Other Body Fluid: Submit urine, CSF, BAL and all other body fluids in a sterile screw cap container.STABILITY Room Temperature: 2 weeks Refrigerated: 2 weeks Frozen: Indefinitely	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22
Bloc surrénalien (Prégnénolone, 17-OH prégnénolone, 17-OH progestérone et 11-désoxycortisol)				CHUS	30077 C Bio-Hormone; Rapport Supra 25,0	LCMS		
Bone Marrow Failure Syndrome Panel [HE0801]	1427	135 genes	Lavender Top (EDTA) [2 tubes]	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
BP 180 and BP230	750		Serum	Immunodermatology Laboratory Department of Dermatology 30 North 1900 East, 4A330 SOM Salt Lake City, Utah 84132 Email: immunoderm@hsc.utah.edu		ELISA	$132.32 US	14-Sep-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Brain-Iron Accumulation NGS Panel	11213	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, GTPBP2, PANK2, PLA2G6, SCP2, WDR45	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		26-Mar-25
Breast and Gynaecological Cancer Specific Panel	227		Lavender Top (EDTA)	Invitae (www.invitae.com)	This is a special panel from the Cedars cancer Centre.	Next-Generation sequencing	Available at MUHC.	10-Jul-23
Breast Cancer Panel (Invitae) [01202]	227	ATM (), BARD1 (), BRCA1 (113705), BRCA2 (600185); BRIP1(), CDH1 (192090); CHEK2 (), NBN (), NF1 (), PALB2 (601728) PTEN (601728), RAD50 () STK11 (602216), TP53 (191170)	Lavender Top (EDTA)	Invitae (www.invitae.com)	Breast-Ovarian Cancer, Familial, type 1 (604370); Breast-Ovarian Cancer, Familial, type 2 (612555); Ovarian carcinoma, somatic (167000); Fanconi anemia, complementation group N (610832); PTEN hamartoma tumor syndrome (_); Pancreatic cancer (260350); Peutz-Jeghers syndrome (175200); Breast cancer (114480)	Next-Generation sequencing	Available at MUHC.	10-Jul-23
Brivaracetam [0718SP]	1465		Red Top or Lavender Top only. Store and send serum/plasma frozen.	NMS laboratories		LC-MS/MS		17-Mar-22
Bromine - Total Blood [FBROM]	1465		Royal Blue (EDTA) top tube	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ICP/MS	INSPQ?? TO BE VERIFIED - NO	30-Apr-21
Bullous Autoimmune Skin Disease Panel: Anti-BP180. BP230, Desmoglein-1, Desmpglein-3	750		Serum gel (Gold top)	Mitogen Advanced Diagnostics	BP180, BP230, Desmoglein 1, Desmoglein 3	CBA		13-Jan-26

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
C1 Esterase Inhibitor Panel [5288176]	1402		Serum. Stable 2 h at RT. Freeze immediately. Stable 2 weeks at -20°C.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org) "		ELISA		30-Sep-25
C1q [9000022]	1402		Serum. Stable 4 h at RT. Store frozen.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)		RID		30-Sep-25
C3 Glomerulopathy Complement Panel [C3G-CP]	792		2 mL serum RED TOP ONLY + 2 mL frozen EDTA plasma. Store frozen.	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	DDD & C3GN	serology		1-Mar-25
C3 Nephritic Factor	792		1 ml frozen serum	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)	Dense Deposit Disease (DDD, aka Membranoproliferative Glomerulonephritis Type II, MPGNII)		Available as Facteur C3 nephritique (activité) at CHUQ (HEJ)	6-Oct-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
C3 Nephritic Factor [C3nef C3CSA]	792		1 ml frozen serum	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	Dense Deposit Disease (DDD, aka Membranoproliferative Glomerulonephritis Type II, MPGNII)	1. Immunofixation electrophoresis (IFE) combines the techniques of electrophoresis with immunofixation to detect C3 degradation products, an indirect measure of dysregulation of C3 convertase. Conversion of C3 to C3c is quantitated (Koch, et al., 1981). 2. C3 Convertase Stabilizing Assay (C3CSA) measures the ability of C3Nefs to stabilize C3 convertase on sheep erythrocytes. 3. C3 Convertase Stabilizing Assay with Properdin (C3CSAP) measures the ability of C3Nefs to stabilize C3	Temporarily not available as Facteur C3 nephritique (activité) at CHUQ (HEJ). Preferred over CCCH.	1-Mar-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
C3a [7453041]	792		1 ml frozen EDTA plasma. Whole blood: 2 hours at room temperature Processed plasma: 6 months at -70°C	CBDI Hemostasis & Thrombosis Lab. Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org) . Ph 513-803-3503				6-Oct-22
C5a [7453044]	792		1 ml frozen EDTA plasma. Whole blood: 2 hours at room temperature Processed plasma: 6 months at -70°C	CBDI Hemostasis & Thrombosis Lab. Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org) . Ph 513-803-3503				6-Oct-22
C9orf72 with reflex to SOD1, ATXN2, FUS, & TARDBP [13039]	853	C9orf72 (614260); SOD1 (147450)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Amyotrphic lateral sclerosis		If C9orf72 is only performed, then the price is only $250 USD; Transferred to MUHC	4-Oct-23
CACNB4 Full Gene Sequencing Analysis [MOL227]		CACNB4 (601949)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Episodic ataxia, type 5 (613855)	Sanger sequencing		14-May-20
Calpain 3 DNA Sequencing Test [563]		CAPN3 (114240)	Lavender Top (EDTA)	Athena Diagnostics (www.athenadiagnostics.com)	LGMD2A (253600), Calpainopathy			
Calprotectine fécale [30536]			random stool	L'Hôpital Maisonneuve-Rosement				
CAMURATI-ENGELMANN DISEASE (CED) VIA THE TGFB1 GENE [787]		TGFB1 (190180)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Camurati-Engelmann Disease (131300)	Sanger sequencing		10-Jul-23
CAR Panel by Immunoblot and Immunohistochemistry [CARP]	720		Serum. Store at 4°C. Satble for 7 d only. DO NOT FREEZE.	Ocular Immunology Laboratory, OHSU Biomedical Research Building, Room 253 3181 SW Sam Jackson Park Road Portland, OR 97239, USA				17-Sep-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum [CDG]	406		Red Top. Store serum frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Affinity Chromatography-Mass Spectrometry	This is a confirmatory test following the screening test for CDT testing at the MUHC.	16-Dec-20
Carbohydrate, Urine [CHOU]	1572		Random urine. Store frozen. Stable 21 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Replaces "reducing substances". No longer available at HSJ.	TLC		31-Oct-20
Cardio-Facio-Cutaneous Syndrome NGS Panel (5 genes)		BRAF (164757), KRAS (190070), MAP2K1 (176872), MAP2K2 (601263), SOS1 (182530)	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)	Cardiofaciocutaneous syndrome (115150); Cardiofaciocutaneous syndrome 2 (615278); Cardiofaciocutaneous syndrome 3(615279); Cardiofaciocutaneous syndrome 4 (615280); Fibromatosis, gingival (135300)	NextGen Sequencing		3/26/2025
Cathartic Laxatives Profile, Stool [FCLPS]			Stool	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Mg, Pi			20-Oct-20
CAVEOLINOPATHY TESTING VIA THE CAV3 GENE [467]		CAV3 (601253)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Muscular Dystrophy, Limb-Girdle, Type 1C (607801)	Sanger sequencing		10-Jul-23
CBFB::MYH11 Inv(16) or t(16;16)	1395		Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children				6-May-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Central Hypoventilation And Apnea Panel [PU0401]	775	CHAT, CHRNA1,CHRNB1, CHRND, CHRNE, COLQ, EDN3, GLRA1, MEP2, PHOX2B, PAPSN, RET, SCN4A, SLC6A5, ZEB2	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		Next-Generation sequencing		26-Sep-18
CEREBRAL CAVERNOUS MALFORMATIONS PANEL [1943]	303	KRIT (604214), CCM2 (607292) , PCCD10 (609118)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral Cavernous Malformations 1(116860), Cerebral Cavernous Malformations 2 (603284), Cerebral Cavernous Malformations 3 (603285)	Sequencing and CNV Detection via NextGen Sequencing using PG-Select Capture Probes		10-Jul-23
Cerebral Cavernous Malformations via the CCM2 Gene [6971]	303	CCM2 (607929)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral calvernous malformations-2	NGS		10-Jul-23
Cerebral Cavernous Malformations via the KRIT1/CCM1 Gene [8161]	303	KRIT1 (604241)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral calvernous malformations-1 (116860)	NGS		10-Jul-23
Cerebral Cavernous Malformations via the PDCD10/CCM3 Gene [8815]	303	PDCD10 (609118)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral calvernous malformation-3	NGS		10-Jul-23
Chicken Feather (e85) IgE, Serum	1419		Red Top; Gold SST	In-Common Laboratories	Available at CHUQ (poulet/plumes)	CIEP	ON HOLD	7-Oct-19

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Cholestasis Panel	400	ABCB11 MYO5B* NOTCH2* NR 21 27 NPHP1 PEX1 PEX10 PEX12 PEX2 PEX26 PEX5 PEX6 SERPINA1 SLC25A13 SLC26A3 SMPD1 SPINT2 TJP2 TMEM216 TRMU L TTC37 UGT1A1 VIPAS39 VPS33B ABCB4 ABCC2 AKR1D1 ATP8B1 BAAT CFTR CREB3L3 CYP7B1 DCDC2 DGUOK EPCAM 80 FAH HSD3B7 JAG1 LCT	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-May-19
Choreoacanthocytosis (200150)		VPS13A (605978)	Lavender Top (EDTA)	North York General		2 mutations		
Chronic Granulomatous Disease NextGen Sequencing (NGS) Panel [1971]		CYBA (608508); CYBB (300481); NCF2 (608515); NCF4 (601488)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Chronic Inflammatory Demyelinating Polyneuropathy (CIDP) Test Panel - Nodal and Paranodal Antibodies	336	Neurofascin 140 (NF140), Neurofascin 186 (NF186); Contactin-1 (CNTN1), Contactin-associated protein 1 (CASPR1), and Neurofascin 155 (NF155)	Serum ONLY. Store at -20°C.	BC Neuroimmunology (bcneuro.ca)		CBA fixed	Replaces contactin and neurofascin tesing at Washington U.	18-Apr-24
Chronic Pancreatitis NGS Panel	402	CASR, CFTR, CTRC, PRSS1, SPINK1	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
Chyluia Screen, Random Urine [CSU]	1465		First morning random urine. Store frozen. Stable 10 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		elecrophoresis		13-Sep-21
CLCN1 Full Gene sequencing Analysis [MOL355]; Paramyotonia Congenita (168300)	607	CLCN1 (118425)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Paramyotonia Congenita (168300)	Sanger sequencing		14-May-20
clonoSEQ	1084		Lavender Top (EDTA)	Adaptive Biotechnologies (https://ous.clonoseq.com)	Minimal Residual Disease	Next-Generation sequencing		14-Sep-24
Coccidiodies Antibody by Immunodiffusion [320]	493		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. SABILITY Refrigerated: 14 days Frozen: 6 months	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22
Coccidiodies Antibody IgG, IgM EIA [325]	493		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. CSF: Sterile transport tube STABILITY Refrigerated: 14 days Frozen: 14 days	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Coccidioides Antibody, Serum [SCOC]	493		Gold SST	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	coccidioidomycosis	Complement fixation and immunodiffusion	Refer to Microbiology.	29-Oct-20
Coccidioides Quantitative EIA [315]	493		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. Plasma: Collect plasma specimens in an EDTA, heparin or sodium citrate tube. Centrifuge for 15 minutes and pipette plasma into a plastic screw cap vial. Urine/CSF/BAL/Other Body Fluids: Submit urine, CSF, BAL, and all other body fluids in a sterile screw cap container.STABILITY Room Temperature: 14 days Refrigerated: 14 days Frozen: Indefinitely	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22
COCKAYNE SYNDROME VIA THE ERCC6 GENE [1008]		ERCC6 (609413)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cockayne Syndrome, Type B (133540)	NGS		Ju;y 10 2023
Collagen Type II Antibodies [FFTYC]	1399		Red Top; Gold SST. Stable for 7 d at 4°C or longer at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Enzyme Linked Immunosorbent Assay (ELISA)		15-May-19
Collagen VII IgG Antibody Level	1399		Red Top or Gold SST. Stability Ambient: 7 days Refrigerated: 14 days Frozen: Indefinitely	Immunodermatology Laboratory Department of Dermatology 30 North 1900 East, 4A330 SOM Salt Lake City, Utah 84132 Email: immunoderm@hsc.utah.edu		Enzyme Linked Immunosorbent Assay (ELISA)		6-Jul-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Colon Cancer NGS Panel	231	APC, AXIN2, BMPR1A, BUB1B, CDH1, CDKN2A, CHEK2, EPCAM, EXO1, FLCN, GALNT12, MLH1, MSH2, MSH6, MUTYH, PMS1, PMS2, PTEN, SMAD4, STK11, TP53	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
Colorectal cancer (Li-Fraumeni syndrome)	231	TP53 (191170)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Colorectal cancer (114500); Li-Fraumeni syndrome (151623)	1. Sanger sequencing 2. gene dosage		
ColoVantage (Methylated Septin 9) [16983]	1395	SEPT9	Plasma (EDTA); handle at 4°C; freeze plasma in plastic container. Minimum 10 mL.	Quest Diagnostics Chantilly 14225 Newbrook Dr. Chantilly, VA 20153-0841				11-Apr-18
Common Variable Immunodeficiency Panel (Inviate) [08112]	1042	CD27, CR2, CTLA4, ICOS, IL21, IL21R, LRBA, NFKB2, PIK3CD, PIK3R1, PLCG2, PRKCD, RAC2, STAT3, TNFRSF13B, TNFRSF13C, TNFSF12	Lavender Top (EDTA)	Invitae (www.invitae.com)		Next-Generation sequencing		10-Jul-23
Complement C1q, Quantitative [016824]	1402		Serum. Store at -20°C. Stable 14 d.	LabCorp/Dynacare (https://www.labcorp.com/)				7-Oct-24
Complement C1q,, Serum [C1Q]	1402		Serum. Store and send frozen Stable 28 d. If the specimen thaws, it is unsuitable for analysis.	Mayo Clinical Laboratories, now available at CHUQ		Nepholometry	Available at CHUQ (H lenfant jesus). Tempoarily Unavailaable	30-Sep-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Complement System Disorder Panel Plus Anlaysis [IM0701]	803	ADIPOQ Complement system AD/AR 2 8 ADIPOR1* Complement system AD/AR 4 ADIPOR2 Complement system AD/AR ARMC4* Ciliary dyskinesia AR 13 15 C1QA C1q deficiency AR 2 7 C1QB C1q deficiency AR 4 7 C1QBP Primary immunodeficiency AD/AR 6 C1QC C1q deficiency AR 4 7 C1R Immunodeficiency AD/AR 14 16 C1S Complement component C1s deficiency AR 4 8 C2* Complement component 2 deficiency AR 4 6 C3 Hemolytic uremic syndrome, atypical, Complement component 3 deficiency AD/AR 5 82 C3AR1 Complement system AD/AR 1 3 C4A* Blood group, Chido/Rodgers system BG 1 5 C4B* Complement component 4B deficiency AR 8 C4BPA Complement system AD/AR 2 C4BPB Complement system AD/AR C5 Eculizumab, poor response to, Complement component 5 deficiency AD/AR 5 17 C5AR1 Complement system AD/AR C5AR2 Complement system AD/AR 2 C6 Complement component 6 deficiency AR 7 11 C7 Complement component 7 deficiency AR 14 29 C8A Complement component 8 deficiency AR 2 5 C8B Complement component 8 deficiency AR 7 7 C8G Immunodeficiency AD/AR C9 Complement component 9 deficiency AR 7 7 CCDC39 Ciliary dyskinesia AR 16 38 CCDC40 Ciliary dyskinesia AR 19 32 CCDC65 Ciliary dyskinesia AR 1 CCDC103 Ciliary dyskinesia AR 3 4 CCDC114 Ciliary dyskinesia AR 6 7 CCNO Ciliary dyskinesia AR 9 9 CD46* Hemolytic uremic syndrome, atypical AD/AR 4 64 CD55 Blood group, Cromer system BG 7 6 CD59 CD59 deficiency AR 3 6	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/				2-Feb-18
Comprehensive Ataxia Repeat Expansion Panel (SCA 1, 2, 3, 6, 7, 8, 10, 11, 12, 17, 36, DRPLA & FRDA) [MOL391]	584		Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		11-Nov-24
Comprehensive Cardiovascular NGS Panel	286		Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)				3/26/2025

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Comprehensive Cellular Energetics Defects (NextGen Sequencing Panel and Copy Number Analysis + mtDNA) [NGS301]	636	AARS2 (612035), ABCB7 (300135), ABCB8 (605464), ABCD1 (300371), ABCF2 (612510), ABHD5 (604780), ACAA1 (604054), ACACA (200350), ACACB (601557), ACAD8 (604773), ACAD9 (611103), ACADL (609576), ACADM (607008), ACADS (606885), ACADSB (600301), ACADVL (609575), ACAT1 (607809), ACAT2 (100678), ACLY (108728), ACO1 (100880), ACO2 (100850), ACOX1 (609751), ACOX2 (601641), ACP6 (611471), ACSBG1 (614362), ACSF3 (614245), ACSL1 (152425), ACSL3 (602371), ACSL4 (300157), ACSM1 (614357), ACSM2B (614359), ACSM3 (145505), ADCK3 (606980), ADHFE1 (611083), AFG3L2 (604581), AGK (610345), AGL (610860), AIFM1 (300169), AK2 (103020), AK3 (609290), ALAD (125270), ALAS2 (301300), ALDH3A2 (609523), ALDOA (103850), ALDOB (612724), ALDOC (103870), ANO10 (613728), APOPT1 (616003), APTX (606350), ARMS2 (611313), ARX (300382), ATAD3B (612317), ATP5A1 (164360), ATP5B (102910), ATP5E (606153), ATP7B (606882), ATPAF2 (608918), AUH (600529), BAAT (602938), BAX	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next-Generation sequencing		14-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Comprehensive Dystonia (NGS Panel and Copy Number + mtDNA) [NGS358]	1096	AARS AARS2 AASS ABCA7 ABCD1 ABCD4 ABHD12 ACOX1 ACP2 ACP5 ACTB ACVR1 ADAMTS13 ADAR ADCY5 ADD3 ADH1C ADRA2B AFG3L2 ALDH6A1 ALS2 ANO3 AP1S2 AP3B2 AP3D1 AP4B1 APTX ARHGAP31 ARSA ARV1 ARX ATCAY ATM ATP13A2 ATP1A2 ATP1A3 ATP2B3 ATP7B ATR AUH B4GALNT1 BCAP31 BCL11B BCS1L BICD2 BRAT1 BSCL2 C11orf73 C19orf12 CA2 CACNA1A CACNA1B CACNA1D CARS2 CHMP2B CIT CKAP2L CLPB COASY COL4A1 COL6A3 COQ9 COX10 COX15 CP CRLF1 CTC1 CYB5R3 CYP27A1 DAG1 DCAF17 DDC DDX3X DLAT DLD DMXL2 DNAJC12 DNAJC6 DOCK6 DPYS DRD5 DYNC1H1 EARS2 ECHS1 ECM1 ELAC2 ELP2 EMC1 ERCC6 ERCC8 ERLIN1 FA2H FAR1 FASTKD2 FBXL4 FBXO7 FGFR1 FOXG1 FOXRED1 FRRS1L FTL GAMT GBA GCDH GCH1 GJA1 GJC2 GLB1 GLUD2 GLYCTK GM2A GNA11 GNAL GNAO1 GNAS GPR88 GRIK2 GTF2E2 GTF2H5 HACE1 HIBCH HNRNPH2 HPCA HPRT1 HTRA2 HTT IFIH1 IFNG ISG15 JAM3 JPH3 KCNJ10 KCNMA1 KCNQ2 KCTD17 KMT2B KRIT1 L2HGDH LIPT2 MAPT MARS2 MAT1A MCCC2 MCOLN1 MDH2 MECP2 MECR MFF MGP MICU1 MMADHC MORC2 MPV17 MRE11A MRPS34 MTO1 NADK2 NALCN NDUFA10 NDUFA12 NDUFA2 NDUFA9 NDUFAF2 NDUFAF6 NDUFS3 NDUFS4 NDUFS7 NDUFS8 NHP2 NKX2-1 NME1 NPC1 NPC2 NUP62 OBFC1 OCLN PAH PANK2 PARK2 PCCA PCCB PDE10A PDE8B PDGFB PDGFRB PDHA1 PDHX PET100 PINK1 PLA2G6 PLEKHG2 PLP1 PNKD PNKP PNP PNPLA8 PNPT1 POLR1C POLR3A PPP1R15B PRKCG PRKRA PRRT2 PSEN1 PSMB8 PTEN PTS QDPR RAB18 RBBP8 RLIM RNASEH1 RNASEH2A RNASEH2B RNASEH2C RNASET2 RNF125 RNF216 SAMHD1 SCP2 SDHA SDHAF1 SDHD SERAC1 SETX SEXN4 SGCE SLC19A3 SLC20A2	Lavender Top (2 x 4 mL)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		14-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Comprehensive Epilepsy (NGS Panel + Copy Number Analyis + mtDNA) [NGS385]	1031	ABAT (137150), ABCC8 (600509), ACADSB (600301), ACSF3 (614245), ADSL (608222), ALDH5A1 (610045), ALDH7A1 (107323), ALG11 (613666), AMT (238310), ARHGEF15 (608504), ARHGEF9 (300429), ARX (300382), ASAH1 (613468), ATIC (601731), ATP6AP2 (300556), AUTS2 (607270), BOLA3 (613183), C10ORF2 (606075), CACNA1A (601011), CACNA1G (604065), CACNA1H (607904), CACNA2D2 (607082), CACNB4 (601949), CACNG3 (606403), CADPS2 (609978), CDKL5 (300203), CERS1 (606919), CHD2 (602119), CHRNA2 (118502), CHRNA4 (118504), CHRNB2 (118507), CLCN2 (600570), CLCN4 (302910), CLN3 (607042), CLN5 (608102), CLN6 (606725), CLN8 (607837), CNTNAP2 (604569), COG7 (606978), COG8 (606979), COL6A2 (120240), COQ2 (609825), COQ9 (612837), CPA6 (609562), CSTB (601145), CTSD (116840), D2HGDH (609186), DEPDC5 (614191), DNAJC5 (611203), DNM1 (602377), DOCK7 (615730), DPAGT1 (191350), DYRK1A (600855), EEF1A2 (602959), EFHC1 (608815), EPM2A (607566), ETHE1	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing	Alternative: Fulgent Epilepsy NGS Panel	28-Jul-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Comprehensive Epilepsy NGS Panel	1031	ABAT, ABCB1, ABCC8, ACY1, ADAR, ADCK3, ADGRG1, ADGRV1, ADSL, AGA, AHI1, AKT3, ALDH4A1, ALDH5A1, ALDH7A1, ALG1, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMT, ANK3, APTX, ARFGEF2, ARG1, ARHGEF15, ARHGEF9, ARL13B, ARSA, ARSB, ARX, ASNS, ASPA, ASPM, ATIC, ATP13A2, ATP1A2, ATP2A2, ATP6AP2, ATP6V0A2, ATPAF2, ATR, ATRX, AUH, B4GALT1, BCKDK, BCS1L, BOLA3, BRAF, BRAT1, BRD2, BTD, BUB1B, C10orf2, C12orf57, C12orf65, CACNA1A, CACNA1H, CACNA2D2, CACNB4, CASK, CASR, CBL, CC2D2A, CCDC88C, CCL2, CDK5RAP2, CDKL5, CDON, CELSR1, CENPJ, CEP152, CEP290, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLCN4, CLCNKA, CLCNKB, CLN3, CLN5, CLN6, CLN8, CNTN2, CNTNAP2,	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
Comprehensive Immune and Cytopenia Panel [IM0902]	1076		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		22-Aug-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Comprehensive In-House Testing Panel [OLD NAME: Autoimmune Encephalitis/ Paraneoplastic Full Panel Evaluation, 20 antigens (AEPP_20 Ag)]	1417	NMDAR, GABAB, DPPX, LGI1, CASPR2, AMPAR, CV2,PNMA2, Yo, Hu, Recoverin, SOX1,.Zic4, GAD65, GluR1, GluR5,GlyR, KLH11, GFAP, and IgLON5	Serum/CSF	BC Neuroimmunology (bcneuro.ca)		Screening with Euroimmun mosaic/paraneoplastic immunoblot/in-house rat-brain immunohistochemistry/ immunofluorescence reflex to in-house CBAs	$1,000.00	4-Feb-25
Comprehensive Leukodystrophy/Leukoencephalopathy (NGS Panel and Copy Number Analysis + mtDNA) [NGS372]	964		Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next-Generation sequencing		29-Sep-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Comprehensive Metabolism NGS Panel	1891	ABCC8, ABCD1, ABCD4, ACAA1, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACAT1, ACOX1, ACSF3, ACY1, ADAMTSL2, ADAR, ADSL, AGA, AGK, AGL, AGPAT2, AKT2, ALAD, ALAS2, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMT, ANO10, ANTXR2, APTX, ARG1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATP13A2, ATP6V0A2, AUH, B3GLCT, B4GALT1, BCKDHA, BCKDHB, BSCL2, BTD, C12orf65, CA2, CACNA1S, CAV1, CAV3, CAVIN1, CBS, CD320, CKMT1A, CKMT1B, CKMT2, CLCN1, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CNNM1, CNNM2, CNNM4, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COQ2, COQ6, COQ8A, COQ9, CPOX, CPS1, CPT1A, CPT1B, CPT2, CTNS, CTSA, CTSC, CTSD, CTSK, DBT, DDOST, DGUOK, DHCR7, DHDDS, DLD, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DYM, ECHS1, EGF, ENO3, EPM2A, ETFA, ETFB, ETFDH, FAH, FBP1, FBXL4, FECH, FH, FLNA, FLNB, FOLR1, FUCA1	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Comprehensive Muscular Dystrophy/Myopathy (NextGen Sequencing Panel and Copy Number Analysis + mtDNA) [NGS330]	636	AAAS (605378), AARS (601065), AARS2 (612035), ABCC8 (600509), ABHD5 (604780), ACACA (200350), ACAD9 (611103), ACADS (606885), ACADVL (609575), ACTA1 (102610), ACY1 (104620), ADCK3 (606980), ADSSL1 (612498), AFG3L2 (604581), AGK (610345), AGL (610860), AGRN (103320), AIFM1 (300169), AIMP1 (603605), AIP (605555), ALAD (125270), ALDH18A1 (138250), ALDOA (103850), ALG2 (607905), ALS2 (606352), AMMECR1 (300195), AMPD1 (102770), ANO5 (608662), AP3B2 (602166), AP3D1 (607246), KIAA0415 (613653), APOA1 (107680), APOPT1 (616003), APTX (606350), AR (313700), ARSA (607574), ASAH1 (613468), ASXL2 (612991), ATL1 (606439), ATP1A3 (182350), ATP2A1 (108730), ATP7A (300011), B2M (109700), B3GALNT2 (610194), B3GNT1 (605517), B4GALT1 (137060), BAG3 (603883), BCL11B (606558), BCS1L (603647), BICD2 (609797), BIN1 (601248), BSCL2 (606158), C10ORF2 (606075), C12orf65 (613541), C19orf12 (614297), C9orf72 (614260), CACNA1A (601011), CACNA1S (114208), CAPN3 (114240), CASK (300172), CASQ1 (114250), CAV3 (601253), CCDC78 (614666), CCDC88A (609736), CFL2 (601443), CHAT (118490), CHCHD10 (615903), CHD4 (603277), CHKB (612395), CHMP2B (609512), CHRNA1 (100690), CHRNB1 (100710), CHRND (100720), CHRNE (100725), CHRNG (100730), CIT (605629), CLCN1 (118425), CLCNKB (602023), CNBP (116955), CNTN1 (600016), COL4A1 (120130), COL6A1 (120220), COL6A2 (120240), COL6A3 (120250), COLQ (603033), COQ2 (609825), COQ9 (612837), COX10 (602125), C12ORF62 (614478), COX15 (603646), FAM36A (614698), COX6A1 (602072), COX6B1 (124089), COX8A (123870), CPT1C (608846), CPT2 (600650), CRYAB (123590), CTNS (606272), CYP7B1 (603711), D2HGDH (609186), DARS2 (610956), DCPS (610534), DCTN1 (601143), DDHD1	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Includes Limb Girdle Muscular Dystrophy	Next-Generation sequencing	Transfered to HSJ.	8-Nov-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Comprehensive Neuropathies (NextGen Sequencing Panel and Copy Number Analysis; 869 genes + mtDNA) [NGS445]	643 (445)	43717 AAAS AARS AASS ABAT ABCA1 ABCB7 ABCC8 ABCD1 ABCD4 ABHD12 ACO2 ACOX1 ACP5 ACTA1 ACTB ACVR1 ACVRL1 ACY1 ADAMTS2 ADCK3 ADCY5 ADCY6 ADSL ADSSL1 AFG3L2 AGXT AIFM1 AIMP1 AKT1 ALAD ALDH18A1 ALDH3A2 ALDH5A1 ALDH6A1 ALG2 ALOX5AP ALS2 AMACR AMER1 AMN AMPD1 AMPD2 ANG ANKH ANKLE2 ANO5 AP1S1 AP1S2 AP3D1 AP4B1 AP4E1 AP4M1 AP4S1 AP5Z1 APOA1BP APOB APTX AR ARCN1 ARHGEF10 ARL13B ARL6IP1 ARNT2 ARSA ARX ASAH1 ASCC1 ASNS ASPA ASXL1 ATAD3A ATL1 ATL3 ATP13A2 ATP1A3 ATP2A1 ATP2B3 ATP6AP2 ATP7A ATP7B ATRX AUH B3GALT6 B3GALTL B4GALNT1 BAG3 BCAP31 BCOR BCS1L BICD2 BIN1 BMP2 BOLA3 BRAT1 BRPF1 BSCL2 BTNL2 BUB1B C10ORF2 C11orf73 C12orf65 C19orf12 CACNA1A CACNA1D CACNA1G CAPN1 CAPN3 CARS2 CASK CAV1 CAV3 CCDC88C CCND1 CCT5 CD59 CDK5 CECR1 CEP120 CEP55 CFH CFHR1 CFHR3 CFL2 CHCHD10 CHMP1A CHMP2B CHRNA1 CHRND CHRNG CISD2 CIT CKAP2L CLCF1 CLCN1 CLCN7 CLP1 CLPB CLPP CNBP CNTN2 CNTNAP1 COASY COG2 COL18A1 COL4A2 COL6A1 COL6A2 COL6A3 COMP COQ2 COQ4 COQ7 COQ9 COX10 COX15 COX6A1 COX7B CPOX CPT1C CREBBP CRYAB CSF1R CSPP1 CTC1 CTDP1 CTNNB1 CTSD CTSF CUBN CWF19L1 CYB5R3 CYP27A1 CYP2U1 CYP7B1 D2HGDH DARS DARS2 DCAF8 DCTN1 DDC DDHD1 DDHD2 DDOST DDR2 DDX3X DES DGUOK DHCR24 DHFR DHH DHTKD1 DKC1 DLAT DMXL2 DNA2 DNAJB2 DNAJC12 DNAJC3 DNM1L DNM2 DNMT1 DOCK6 DPAGT1 DPH1 DPM1 DSE DST DYNC1H1 EARS2 EBF3 ECE1 EGR2 EIF2B1 EIF2B2 EIF2B3 EIF2B4 EIF2B5 ELOVL4 ELOVL5 ELP2 EMC1 EML1 ENTPD1 EPG5 ERBB3 ERBB4 ERCC1 ERCC2 ERCC3 ERCC6 ERCC8 ERLIN1 ERLIN2	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		14-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Comprehensive Ophthalmoplegia Syndromes (NextGen Sequencing Panel and Copy Number Analysis; 55 Genes + mtDNA) [NGS352]	1648	ACADS (606885), ACTA1 (102610), AFG3L2 (604581), AGRN (103320), APTX (606350), ATXN1 (601556), ATXN2 (601517), ATXN3 (607047), ATXN7 (607640), BIN1 (601248), C10ORF2 (606075), C12orf65 (613541), CHAT (118490), CHRNA1 (100690), CHRNB1 (100710), CHRND (100720), CHRNE (100725), CLPP (601119), COLQ (603033), DNM2 (602378), DOK7 (610285), EARS2 (612799), FOXE3 (601094), FOXRED1 (613622), GBA (606463), GRM1 (614831), HARS2 (600783), HCCS (300056), HSD17B4 (601860), KIF21A (608283), LARS2 (604544), C20ORF72 (615076), MPV17 (137960), MTM1 (300415), MTMR14 (611089), MUSK (601296), MYH2 (160740), OPA1 (605290), OPA3 (606580), PABPN1 (602279), POLG (174763), POLG2 (604983), RAPSN (601592), RRM2B (604712), RYR1 (180901), SCO2 (604272), SDHAF1	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		14-May-20
Comprehensive Short Stature Syndrome Panel [MA2101]	560		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		Sanger sequencing		25-Sep-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Cone-Rod Dystrophy Panel [10403]	1444	CACNA1F (300110)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Aland Island Eye Disease (300600); Cone-Rod Dystrophy X-Linked 3 (300476); Congenital Stationary Night Blindness, Type 2A (300071)	NextGen Sequencing	See Blueprint Genetics	10-Jul-23
Congenital Adrenal Hyperplasia NGS Panel	1326	ARMC5, CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD3B2, POR, PRKAR1A, STAR	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)				3/26/2025
Congenital Adrenal Hyperplasia Panel [EN0801]	1326	ARMC5 CYP11A1 CYP11B1* CYP11B2* CYP17A1 CYP21A2* HSBD3B2 PDE11A PDE8B POR PRKAR1A STAR	Lavender Top (EDTA) 2 x 4 mL	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Congenital Hyperinsulinism Panel [10265]	541	ABCC8 (600509) GCK (138079) GLUD1 (138130) HADH (601609) HNF1A (142410) HNF4A (600281) KCNJ11 (600937) SLC16A1 (600682) UCP2 (601693)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hyperinsulinemic Hypoglycemia, Familial 3 602485 Hyperinsulinemic Hypoglycemia, Familial 6 606762 Hyperinsulinemic Hypoglycemia, Familial, 1 256450 Hyperinsulinemic Hypoglycemia, Familial, 2 601820 Hyperinsulinemic Hypoglycemia, Familial, 4 609975 Hyperinsulinemic Hypoglycemia, Familial, 7 610021 Maturity-Onset Diabetes Of The Young, Type 1 125850 Maturity-Onset Diabetes Of The Young, Type 3 600496 Related Tests	Next-Generation sequencing		7/10/2023

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Congenital Myasthenic Syndrome NGS Panel	606	AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LAMB2, LRP4, MUSK, MYO9A, PLEC, PREPL, RAPSN, SCN4A, SLC25A1, SLC5A7, SNAP25, STIM1, SYT2 (28 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Myasthenic syndrome, congenital, with pre- and postsynaptic defect s 615120); Myasthenic syndrome, congenital, associated with episodic apnea (254210); Multiple pterygium syndrome, lethal type (253290); Myasthenic syndrome, fast-channel congenital (608930); Myasthenic syndrome, slow-channel congenital (601462); Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency (608931); Myasthenic syndrome, slow-channel congenital (601462); Multiple	Next-Generation sequencing		3/26/2025

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Congenital Neutropenia Panel [IM0501]	836	ACTB* CLPB CSF2RA#* CSF3R CTSC EFL1* ELANE G6PC3 GATA2 GFI1 GINS1 HAX1 IFNGR2 JAGN1 LAMTOR2 LYST MKL1 PGM3 RAC2 SBDS* SLC37A4 SMARCD2 SRP54 SRP72* VPS13B VPS45# WAS WDR1	Lavender Top (2 x 4 mL)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Connective Tissue Disease- Lupus Profile, Serum [CTDAB S]	1863	Anti Mi-2 beta Ab (Anti Chromodomain helicase DNA binding protein 4 antibody); Anti Ku Ab; Anti nRNP/Sm Ab (Anti Nuclear ribonucleoprotein (U1-RNP) antibody); Anti Sm Ab (Anti Smith antibody); Anti SSA (Ro60) Ab; Anti Ro52 (TRIM21) Ab; Anti SSB (La) Ab; Anti Scl-70 Ab (Anti Topoisomerase I antibody); Anti PM/Scl-100 Ab (Anti PM/Scl Complex (100 kDa) antibody); Anti Jo-1 Ab (Anti Histidyl-tRNA synthetase antibody); Anti Centromere protein B Ab; Anti PCNA Ab (Anti Proliferating cell nuclear antigen antibody); Anti dsDNA Ab (Anti Double stranded DNA antibody); Anti Nucleosome (chromatin) Ab; Anti Histone Ab; Anti Ribosomal P proteins Ab; Anti AMA-M2 Ab (Anti	Gold SST	In-Common Laboratories		Immunoblot	Replaces Mitogen panel. Mitogen does not accept CSF.	30-Mar-25
Copeptin, Serum/Plasma	1465		Prefered Plasma (Li heparin). Serum & EDTA plasma OK. Store and send frozen (90 days).	In-Common Laboratories	Replaces vasopressin/Anti-Diuretic Hormone. Suggest ordering osmolality at same time.	Fluorescent immunoassay		5-Sep-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Copper (Liver Biopsy)		Normal: 1.16-0.55 µmol/g	Tissue Mimimum: 1.0 mg. Collection tube without anticoagulant (plastic) NO additions. Store and ship at 4°C.	INSPQ (Quebec City)				23-Jun-20
Cornelia de Lange Syndrome		NIPBL (608667); SMC1A (300040); SMC3 (606062); HDAC8 (300269); RAD21 (606462)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Cornelia de Lange syndrome 1 (122470); Cornelia de Lange syndrome 2 (300590); Cornelia de Lange syndrome 3 (610759); Cornelia de Lange syndrome 4 (614701); Cornelia de Lange syndrome 5 (300882)	Next-Generation sequencing		3/26/2025
Corticotropin Releasing Factor (CRF, CRH)			3 ml EDTA plasma should be collected and separated as soon as possible. Plasma should be frozen immediately after separation	Inter Science Institute - 944 West Hyde Park Blvd, Inglewood, CA 90302		RIA		
Cortisol, free [CORTF]			Red Top or Lavender Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				31-Oct-20
Coxiella burnetii (Q fever), Molecular Detection, PCR, Blood [CBBRP]			Whole blood EDTA; Stable 7 days only at at 4°C or -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Fièvre Q (Coxiella burnettii IgG et IgM) at CHUS Fleurimont			31-Oct-20
Craniosynostosis Non-Syndromic (select exons of FGFR3 gene)		FGFR3 (134934)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis	Sanger sequencing; FGFR3 (p.Pro250Arg)		

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Creatine Disorders Panel, Urine [CRDPU]		GATM (602360); SLC6A8 (300036)	Random urine	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	arginine:glycine amidinotransferase deficiency (602360/612718), guanidinoacetate methyltransferase deficiency (601240/612736), creatine transporter (SLC6A8) defect (300036/300352)		To be verified.	
Crouzon Syndrome (select exons of FGFR2 and FGFR3 gene)		FGFR2 (176943); FGFR3 (134934)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis: Crouzon syndrome (123500)	Sanger sequencing: FGFR2 (exon 7 and 8); FGFR3 (p.Pro250Arg)	To be verified.	

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Cryoglobulin and Cryofibrinogen Panel, Serum and Plasma [CRGSP]			**Cryofibrinogen Collection** Container/Tube: Lavender top (EDTA) Submission Container/Tube: Plastic vial Specimen Volume: 1 mL Collection Instructions: 1. Tube must remain at 37 degrees C. 2. Centrifuge at 37 degrees C. (Do not use a refrigerated centrifuge. If absolutely necessary, ambient temperature is acceptable.) It is very important that the specimen remain at 37 degrees C until after separation of plasma from red cells. 3. Place plasma into an appropriately labeled plastic vial. **Cryoglobulin Collection** Container/Tube: Red top Submission Container/Tube: Plastic vial Specimen Volume: 5 mL Collection Instructions: 1. Tube must remain at 37 degrees C. 2. Allow blood to clot at 37 degrees C. 3. Centrifuge at 37 degrees C	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Evaluating patients with vasculitis, glomerulonephritis, and lymphoproliferative diseases Evaluating patients with macroglobulinemia or myeloma in whom symptoms occur with cold exposure	immunofixation	To be verified.	

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Cryopyrin-Associated Periodic Syndromes via the NLRP3 Gene [1638]		NLRP3 (606416)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Chronic Infantile Neurological, Cutaneous And Articular Syndrome (607115); Familial Amyloid Nephropathy With Urticaria And Deafness (191900); Familial Cold Urticaria	Sanger sequencing		10-Jul-23
Cryptococcal Antigen, latex agglutination [319]	2020		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. CSF: Collect in sterile container. Centrifuge to remove white cells and particulate matter. STABILITY efrigerated/Frozen: 2 months	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22
CSF Protein Immunoassay Panel (CJD Protein Test Panel)	1383		2.0 mL CSF; Freeze sample as soon as possible after collection. Ship frozen on dry ice.	National Microbiology Laboratory, Health Canada (Winnipeg)	Creutzfeldt-Jakob Disease (CJD)	14-3-3 protein testing of cerebrospinal fluid (CSF) (ELISA); PrPd (QuIC); Tau protein (ELISA)	Should be registered with surveillance.	24-May-17
CSTB dodecmer repeat expansion [7084]		CSTB (601145)	Lavender Top (EDTA)	Ambry Genetics	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg) (254800)		To be verified.	
Currarino syndrome		MNX1 (142994)	Lavender top (EDTA)	Diagenos (www.diagenos.com)	Currarino syndrome (176450)	Sanger sequencing		

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Currarino syndrome		MNX1 (142994)	Lavender top (EDTA)	Centogene AG (www.centogene.com)	Currarino syndrome (176450)	Sanger sequencing		
CXCL9	1465		EDTA plasma - 2 aliquots. Store in polypropylene tubes.. Send frozen. Satble 6 month	Cincinnati Children's Hospital Medical Center, Diagnostics Immunolgy Laboratory, 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)	hemophagocytic lymphohistiocytosis (HLH)			18-Apr-23
Cyclic AMP, Urinary Excretion [GRP]			Serum (Red Top) AND random urine resquired. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				19-May-17
CYP27B1 Single Gene Test	897	CYP27B1 (609506)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Vitamine D dependant rickets type1 (264700)	NextGen Sequencing		3/26/2025
Cystatin C [CYSTC]	78		Serum. Store and send frozen	In-Common Laboratories		Immunonephelometry		6-Sep-18
Cystic Fibrosis: CFTR Deletion Duplication Analysis	763	CFTR (602421)	Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children (www.sickkids.ca/paediatriclabmedicinems/lab-divisions/genome-diagnostics/genome-diagnostics.html#genome)	Cystic Fibrosis (219700)	MLPA	Available at MUHC	
Cystinuria NGS Panel	807	SLC3A1 (104614); SLC7A9 (604144); PREPL (609577)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Cystinuria (220100)	NextGen Sequencing		3/26/2025
Cytochrome P450 2D6 (CYP2D6) Comprehensive Cascade [2D6CV]	77	CYP2D6 (124030)	Lavender Top (EDTA). Stable for 30 days at 4°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				31-Oct-20
Cytokine Panel [CYPAN]	1523		Lavender Top (EDTA). Stable for 30 days at 4°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	includes soluble interleukin-2 receptor	bead-based multiplex immunoassay	See OncoHelix	18-Apr-23
Dabigratan			Serum (Light blue top)	Quest Diagnostics				

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
DARS2 Full Gene Sequencing Analysis [MOL094]		DARS2 (610956)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation (611105) Alternate name: DARS2: Mitochondrial Aspartyl-tRNA Synthetase Deficiency	Sanger sequencing		14-May-20
Deletion 1p			Green Top; RT only	Cytogenetics Laboratory, Hospital for Sick Children	Chromosome1p36 deletion syndrome (607872)	microarray	To be verified.	
Dementia, Plus APOE Panel [15779]	598	19 genes	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NGS	Replaces Dementia Panel [10309]	20-Jun-24
Developmental Disorders Panel [MS020x]	718		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NGS	Only temporarily allowed. To be replaced by polymalformation Panel in Qc	8-Oct-24
Dexamethasone [FDXM]	1465		Red Top preferred. Serum stable for 28 d at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS		6-Oct-22
Diabetes Insipidus Panel [KI1801]	1457	AQP2 AVP AVPR2	Lavender Top (EDTA) 2 x 4 mL	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
DICER1 single gene test [S00555]	246	DICER1	Lavender Top (EDTA) 2 x 4 mL	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
Dihydrotestosterone, Serum [DHT]	1459		Red Top or Gold Top. Store serum and send frozen	In-Common Laboratories		ELISA		30-Jan-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Disaccharidase Determination, Small Bowel Biopsy	85		Intestinal biopsy, 2-5 mg wet weight. Store at -70°C until shipping and send on dry ice	Gastroenterology Clinical Lab. Nemours/Alfred I.Dupont Hospital for Children, Wilmington, DE (www.nemours.org) (https://www.nemours.org/pediatric-research/labservices/diagnostic/gastroenterology-lab.html)		lactase, maltase, sucrase, palatinase and glucoamylase		22-Feb-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Disorders of Sex Development Sequencing Panel with CNV Detection [4509]	1395	AMH 600957 AMHR2 600956 ANOS1 300836 AR 313700 ARL6 608845 ARX 300382 ATRX 300032 BBS1 209901 BBS10 610148 BBS12 610683 BBS2 606151 BBS4 600374 BBS5 603650 BBS7 607590 BBS9 607968 CBX2 602770 CHD7 608892 CYP11A1 118485 CYP17A1 609300 CYP19A1 107910 DHH 605423 DMRT1 602424 DMRT2 604935 FGF8 600483 FGFR1 136350 FGFR2 176943 FOXL2 605597 FSHB 136530 GATA4 600576 GNRH1 152760 GNRHR 138850 HESX1 601802 HFE 613609 HS6ST1 604846 HSD17B3 605573 KISS1 603286 KISS1R 604161 LEP 164160 LEPR 601007 LHCGR 152790 LHX3 600577 LHX4 602146 MAMLD1 300120 MAP3K1 600982 MKKS 604896 NR0B1 300473 NR5A1 184757 NSMF 608137 PCSK1 162150	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	3-Oxo-5 Alpha-Steroid Delta 4-Dehydrogenase Deficiency AD 264600 46,XX Sex Reversal, Type 1 AR 400045 46,XY Sex Reversal, Type 3 AD 612965 46,XY Sex Reversal, Type 5 AD 613080 46,XY Sex Reversal, Type 6 AD 613762 46,XY Sex Reversal, Type 7 AR 233420 Adrenal Insufficiency, Congenital, With 46,XY Sex Reversal, Partial Or Complete AR 613743 Androgen Resistance Syndrome XL 300068 Antley-Bixler Syndrome AR 207410 Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis AR 201750 ATR-X Syndrome XL 301040 Bardet-Biedl Syndrome 1 AR 209900 Bardet-Biedl Syndrome 10 AR 615987 Bardet-Biedl Syndrome 11 AR 615988 Bardet-Biedl Syndrome 12 AR 615989 Bardet-Biedl Syndrome 2 AR 615981 Bardet-Biedl Syndrome 3 AR 600151 Bardet-Biedl Syndrome 4 AR 615982 Bardet-Biedl Syndrome 5 AR 615983 Bardet-Biedl Syndrome 6 AR 605231 Bardet-Biedl Syndrome 7 AR 615984 Bardet-Biedl Syndrome 8 AR 615985 Bardet-Biedl Syndrome 9 AR 615986 Blepharophimosis, Ptosis, And Epicanthus Inversus AR 110100 Camptomelic Dysplasia AD 114290	Exome sequencing with CNV detection		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Diuretic Screen, Urine [FDIRU]			Random urine	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	benzthiazide, bumetanide, chlorothiazide, chlorthalidone, furosemide, hydrochlorothiazide, hydroflumethiazide, and metolazone		To be verified.	
DNA Double-Standed Antibodies, IgG, Serum [ADNA]	1465		Serum SST. Stable at -20°C for 21 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELISA		20-Aug-21
Double-stranded DNA (dsDNA) Antibodies, Serum/Plasma [DNAAB]	1465	ant-dsDNA	Serum; Plasma (EDTA); Plasma (heparin); Plasma (Citrate). Store at -20°C.	In-Common Laboratories		ELISA	Replaces Mitogen.	31-Mar-25
Doxycycline [94093]	1465		Serum (2 mL). Store frozen.	Quest Diagnostics				21-Feb-20
Drug Dependent Platelet Antibody [9000]	1465		Gold SST	Blood Center of Wisconsin				
Drug Screen, Prescription/OTC, Urine [PDSU]	1465		Random urine	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Trifluoperazine (stelazine)			20-Oct-20
Duchenne and Becker Muscular Dystrophy (310200 and 300376)	617	DMD (300377)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. Sanger sequencing 2. gene dosage		4-May-17

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Dystonia Dyskinesia NGS Panel	1096	ANO3, ATP1A3, CACNA1B, CIZ1, COL6A3, DRD2, DRD5, GCH1, GNAL, HPCA, KCTD17, PNKD, PRKN, PRKRA, PRRT2, SCP2, SGCE, SLC2A1, SLC6A3, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Alternating hemiplegia of childhood 2 (614820); CAPOS syndrome (601338); Dystonia-12 (128235); Dystonia, myoclonic (159900); Dystonia, primary cervical; Dystonia 16 (612067); Convulsions, familial infantile, with paroxysmal choreoathetosis (602066); Episodic kinesigenic dyskinesia 1 (128200); Seizures, benign familial infantile, 2 (605751); Leukoencephalopathy with dystonia and motor neuropathy (613724); Dystonia-11, myoclonic (159900); Parkinsonism-dystonia, infantile (613315); Dystonia, dopa-responsive	Next-Generation sequencing		3/26/2025
Dystonia/Parkinson Panel [T402]	1096		Lavender Top (EDTA)	GeneDx (www.genedx.com)		Next-Generation sequencing		20-Feb-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Early-Onset Epileptic Encephalopathy Panel		ACY1, ADGRV1, ADSL, ALDH7A1, ALG13, ARFGEF2, ARHGEF15, ARHGEF9, ARX, ATP1A2, ATP6AP2, AUH, BCKDK, CACNA1A, CACNA2D2, CACNB4, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CPA6, CSTB, CTSD, CTSF, DEPDC5, DNAJC5, DNM1, DYNC1H1, DYRK1A, EEF1A2, EFHC1, EPM2A, FARS2, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GNAO1, GOSR2, GRIN1, GRIN2A, GRIN2B, HCN1, HCN2, HNRNPU, IQSEC2, KANSL1, KCNB1, KCNH2, KCNH5, KCNJ10, KCNQ2, KCNQ3, KCNT1, KCTD7, KPNA7, LGI1, LIAS, MAGI2, MBD5, MECP2, MEF2C, MFSD8, MTOR, NEDD4L, NHLRC1, NR2F1, NRXN1, PCDH19, PIGA, PIGO, PIGV, PLCB1, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRRT2, QARS, RBFOX1, RBFOX3, ROGDI,	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)				3/26/2025
Eculizumab	909		Serum 0.5 mL. Separate and freeze within 2 h of collection.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)	Solaris	ELISA	Now available at HSJ	22-Aug-18

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Ehlers-Danlos Syndrome Panel [CA0101]	575	ADAMTS2, ATP7A, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, FLNA, PLOD1, SLC39A13	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing; Del Dupl		1-Feb-24
Encephalopathy, Autoimmune Evaluation [ENCES CSF]			CSF minimum 3 cc	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	IFA, RIA, CBA, WB	Requires special permission		9-Aug-22
Encephalopathy, Autoimmune Evaluation [ENCES serum]			Red top, 7-10 cc of blood	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	IFA, RIA, CBA, WB	Requires special permission		9-Aug-22
Epidermolysis bullosa (226600)		COL7A1 (120120)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Essential Epilepsey Panel	356	ACY1, ADSL, ALDH7A1, ALG13, ARHGEF9, ARX, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CNTNAP2, DEPDC5, DNM1, FOLR1, FOXG1, GABRA1, GABRB3, GABRG2, GAMT, GNAO1, GRIN2A, GRIN2B, HCN1, HNRNPU, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNT1, LGI1, MAPK10, MBD5, MECP2, MEF2C, NRXN1, PCDH19, PLCB1, PNKP, PNPO, POLG, PRRT2, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SIK1, SLC19A3, SLC25A22, SLC2A1, SLC35A2, SLC9A6, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYNGAP1, SZT2, TBC1D24, TCF4, TREX1, TSC1, TSC2, UBE3A, ZEB2	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)				3/26/2025
Estrone, Serum [E1]	1465		Red Top Only. Store at -20°C for up to 28 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	disorders of sex steroid metabolism	LC-MS		21-Nov-22
Everolimus, Blood [EVROL]			Lavender Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	TDM		HSJ; whole blood EDTA, store at 4°C	
F-Actin Ab, IgG, Serum [FACT]	1969		Serum (Gold or Red Top). Stable for 21 days at 4°C (preferred) or -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELISA		17-Aug-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Factor B			0.5 mL red top serum- spun, separated, frozen within 2 hrs of collection; ship on dry ice	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)				15-Nov-17
Factor H	64		2ml frozen serum.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)		complement-mediated renal diseases FH (factor H)	Alternate sites Iowa, CHUQ (HEJ), & National Jewish (B1H)	6-Jan-23
Factor H autoantibodies	64		2ml frozen serum.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)			Factor H Autoantibodies (CHU S-J) - plasma	6-Jan-23
Familial Hemophagocytic Lymphohistiocytosis (FHL), Autosomal Recessive; type 2 (603553), type 3 (608898), type 4 (603552) and type 5 (613101)		PRF1 (170280); UNC13D (MUNC13-4) (608897); STXBP2 (601717); RAB27A (603868); STX11 (605014)	Lavender Top (EDTA)	Cinncinatti Children's Hospital (Division of Human Genetics Diagnostic Laboratories)	PRF1 and STX11 also at: Hospital for Sick Children		Available at Fulgent for $1110	
Familial Hypocalciuric Hypercalcemia (FHH) Panel [10125]	805	AP2S1, CASR, GNA11	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Next-Generation sequencing		10-Jul-23
Familial Limb Girdle Myasthenia Syndrome via DOK7 Gene [7629]	626	DOK7 (610285)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Myasthenia, Limb-Girdle, Familial (254300)		Available at MNG	10-Jul-23
Farmer's Lung IgG Antibodies, Serum [FLGAB]	1465		Serum (2 mL). Store frozen.	In-Common Laboratories	M. faeni IgG Ab; T. vulgaris IgG Ab	FEIA		10-Mar-20
Fat, Feces [FATF]	1465		For a random collection, a minimum of 5 g (do not send entire collection) is required. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Nuclear Magnetic Resonance (NMR) Spectroscopy		3-Aug-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Fatty Acid Oxidation Deficiency NGS Panel	528	ACAD9, ACADL, ACADM, ACADS, ACADVL, CPT1A, CPT1B, CPT2, ETFA, ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, SLC22A5, SLC25A20, TAZ	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Includes: Fatty liver, acute, of pregnancy (609016); HELLP syndrome, maternal, of pregnancy (609016)	NextGen Sequencing		3/26/2025
Fatty Acid Oxidation Syndrome Panel [ME1701]	1320		Lavender Top (2 x 4 mL)	Blueprint Genetics http://blueprintgenetics.com/		Next-Generation sequencing		29-Feb-20
FH Autoantibody Testing	64		Panel requires at least 2ml frozen serum.	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	Hemolytic uremic syndrome, atypical, susceptibility to, 5 (612925)	ELSIA and other assays		1-Mar-25
FH Gene Sequencing & Del/Dupl [713]	1034	FH (136850)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	fumarate hydratase; fumarase deficiency. Includes: Hereditary Leiomyomatosis and Renal cell Cancer (HLRCC) (150800)	Sanger sequencing and del/dupl		19-Jan-23
Fibrodysplasia ossificans progressiva (135100)		ACVR1 (102576)	Lavender Top (EDTA)	University of Pennsylvania School of Medicine	Fibrodysplasia ossificans progressiva (135100)	ACVR1 point mutation C617 G-A		
FLT3/NPM1	1395		Lavender Top (EDTA). Bone marrow. Stable 24-48 h at RT after draw. Do not freeze.	Dept of Clinical Laboratory Genetics. Genome Diagnostics. Toronto General Hospital.		RNA	Reminder: Mark Urgent on Requisition	10-May-22
Fluconazole, Serum or Plasma [2089SP]	1118		Serum: Collect sample in Red top tube Plasma: Collect sample in Lavender top tube (EDTA)	NMS Laboratories		Liquid Chromatography/Tandem Mass Spectrometry (LC/MS/MS)		9-Jun-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Fluphenazine (Prolixin), Serum [PROLX]	929		Serum Draw blood in a plain red-top tube(s), serum gel tube is not acceptable. Spin down and send 3 mL of serum refrigerated in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Modecate	Liquid Chromatography/Tandem Mass Spectrometry (LC/MS/MS)		26-Jan-20
Focused Cytokine, Chemokine Growth Factor 15-plex	1523		Collect blood in a plasma EDTA tube (Purple Vacutainer) or in a serum collection tube (Gold (SST) or Red Vacutainer). Store at -20*C. Stable 1 yr at -80°C.	Mitogen Advanced Diagnostics		ALBIA		6-Jan-26
Focused Pharmacogenomics Panel [PGXFP]	1582	CYP1A2, CYP2C9, CYP2C19, CYP2D6, CYP3A4, CYP3A5, SLCO1B1, VKORC1, CYP4F2, and rs12777823	Lavender Top (EDTA); Saliva; DNA	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		TaqMan		24-Jan-25
Friedreich's Ataxia DNA Sequencing Analysis [348]	1710	FXN (FRDA) (606829)	Lavender Top (EDTA)	Athena Diagnostics (www.athenadiagnostics.com)	Friedreich ataxia (229300). Hop. Saint-Justin - triplet expansion; North York - triplet expansion $445	Sanger sequencing	Available at HSJ [55142]	27-Jun-00
FSHD - Detection of Abnormal Alleles with Interpretation (FSHD1 and FSHD2)	102	DUX4 (D4Z4) (606009); SMCHD1 (614982)	Lavender Top (EDTA). Rejection: Specimens collected more than 5 days before receipt by the laboratory.	University of Iowa Diagnostic Laboratories (http://www.healthcare.uiowa.edu/path_handbook/rhandbook/test127.html)	FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1 (158900); Facioscapulohumeral Muscular Dystrophy 2 (158901)	Southern blot, 4qA.4qVhaplotyping; methylation; SMCHD1 sequencing		13-May-19

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
FUNGITEL BETA-D -Glucan Colorimetric Assay [317]	489		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a transport tube without interfering levels of (1→3)-β-D-Glucan. Most sterile polypropylene DNAase and RNAse free tubes are acceptable. CSF: Submit CSF in a sterile screw cap container. STABILITY Room Temperature: Not acceptable. Refrigerated: 5 days Frozen: Indefinitely	MiraVista Diagnostics	Elevated in fungal infection	Colorimetric Fungitell® assay utilizing a (1→3)-β-D-Glucan - specific Limulus Amebocyte Lysate (LAL)	No longer performed in province. AH-612 must be approved by Microbiology.	8-Sep-22
Fungitell, Serum [FUNGS]	489		Collect 3-5 mL blood in a serum separator gel tube (SST). Centrifuge specimen within 2 hours. Ship serum gel tube frozen	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Replaced by: 1,3-Beta-D-Glucan (Fungitell), Serum		20-Jan-20
Gabapentin [GABA]	107		Red Top. Store and send frozen.	In-Common Laboratories		LC/UV		6-Sep-18
Galactose-alpha-1,3-galactose (Alpha-Gal) IgE [ALGAL]	1761		Collect RED or GOLD SST. Store at 4°C for 14 days or at -20°C (90 days).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Phadia ImmunoCAP	Phadia ImmunoCAP	Available at ICL $120	9-May-23
Ganglioside Antibodies Profile IgG & IgM, Serum [GANGM S]	32	Anti GM1 IgG, Anti GM2 IgG, Anti GM3 IgG, Anti GD1a IgG, Anti GD1b IgG, Anti GT1b IgG, Anti GQ1b IgG, Anti GM1 IgM, Anti GM2 IgM, Anti GM3 IgM, Anti GD1a IgM, Anti GD1b	Serum (Red Top); Plasma (EDTA, Heparin, Citrate). Stable 14 d. Store frozen.	In-Common Laboratories		EUROImmun EUROBlotOne	Replaces Mitogen Neurological Disease Profile	9-Mar-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
GARS Full Gene Sequencing Analysis [MOL167]		GARS (600287)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Charcot-Marie-Tooth disease, type 2D (601427); Neuropathy, distal hereditary motor, type V (600794)	Sanger sequencing		9-May-20
Gaucher Disease (recurrent mutations)		GBA (606463)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Gaucher's Disease, Type 1 (230800); Gaucher Disease, Perinatal Lethal (608013); Subacute Neuronopathic Gaucher's Disease (230900); Gaucher Disease, Type IIIc (231005)	Direct mutation analysis (9 mutations);AKJ 90% sensitivity; others 50-60%		
GAUCHER DISEASE VIA THE GBA1/GBA GENE [479]	464	GBA (606463)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Gaucher's Disease, Type 1 (230800); Gaucher Disease, Perinatal Lethal (608013); Subacute Neuronopathic Gaucher's Disease (230900); Gaucher Disease, Type IIIc (231005)	Sanger sequencing		4-Jan-24
Gene dosage for FGFR2, FGFR3 & TWIST		FGFR2 (176943); FGFR3 (134934); TWIST1 (601622)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis	del/dupl	To be verified.	
Genetic Eye Disease Panel for **Strabismus** (Gedi-S)		ROBO3, PHOX2A, HOXA1, SALL4, CHN1, TUBB3, KIF21A, HOXB1	Lavender Top (EDTA) 2-5 cc	Ocular genomics (https://ocular genomics.meei.harvard.edu/index.php/gdt)	Strabismus	NextGen Sequencing	To be verified.	

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Genetic Renal Panel	1285	CFH (134370), CFI (217030), MCP (120920), CFB (138470), CFHR5 (608593), C3 (120700), THBD (188040), ADAMTS13 (604134), DGKE (601440), PLG (173350); CFHR3-CFHR1 (605336/134371)	Lavender Top (EDTA)	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	thrombotic microangiopathies: Hemolytic Uremic Syndrome, atypical Hemolytic Uremic Syndrome and Thrombotic Thrombocytopenic Purpura	Next-Generation sequencing; MLPA		1-Mar-25
Ghrelin	1465		Collect 10mL blood in special **ISI GI Preservative** tube yielding special GI plasma and separate in refrigerated centrifuge as soon as possible. Transfer 3-5mL immediately into non-glass shipping vial. Minimum specimen size is 1mL. Freeze specimen at -20°C.	InterScience Institute		EIA/ELISA		8-Dec-22
Gilbert Syndrome (UGT1A1) Single Gene Test	1340	UGT1A1 (191740)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)	diagnosis of Gilbert (143500) or Crigler-Najjar syndromes (218800 and 606785); irinotecan & others sensitivity	Sanger sequencing		3/26/2025
GLA gene dosage	469	GLA (300644)	Lavender top (EDTA); Store at RT or 4°C for up to 48 h after drawing. At 4°C for >48 h.	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Fabry Disease (301500)	MLPA	To be verified.	

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
GLA gene sequencing	469	GLA (300644)	Lavender top (EDTA); Store at RT or 4°C for up to 48 h after drawing. At 4°C for >48 h.	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Fabry Disease (301500)	Sanger sequencing	To be verified.	
Glucagon, Plasma [GLP]	110		Collection Container/Tube: Lavender top (EDTA) Submission Container/Tube: Plastic vial Specimen Volume: 2 mL Collection Instructions: 1. Fasting. 2. Prechill tube at 4 degrees C before drawing the specimen. 3. Draw the prechilled tube, and process as follows: a. After drawing specimen, chill tube in wet ice for 10 minutes. b. Centrifuge in a refrigerated centrifuge or in chilled centrifuge cup. c. Immediately after centrifugation, remove plasma, place in a plastic transport vial (Supply T465), and freeze.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		RIA		17-Mar-20
GLUCOSE TRANSPORTER TYPE 1 DEFICIENCY SYNDROME (06777)		SLC2A1 (138140)	Lavender Top (EDTA)	BC Children's Hospital & BC Women's Hospital, Canada (http://www.genebc.ca)		Sanger sequencing & reflex MLPA		19-Oct-17

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY VIA THE G6PD GENE [7657]		G6PD (305900)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hemolytic anemia due to G6PD deficiency (300908)	Sanger sequencing		10-Jul-23
GLUT1 Deficiency Syndrome (SLC2A1 Single Gene Test)	262	SLC2A1 (138140)	Lavender Top (EDTA) [2 tubes]	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
Glutamate Receptor R1							NOT AVAILABLE: Wash U, Mayo, Oxford	21-Apr-17
Glycine Receptor Alpha1 IgG, Cell Binding Assay, Serum [GLYCS]	938		Serum. Red Top Only. Store frozen. Stable for 28 days at 4°C and 28 days at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		live cell binding assay		7-Mar-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Glycogen Storage Disease and Disorders of Glucose Metabolism Sequencing Panel with CNV Detection [10385]	476	AGL 610860 ALDOA 103850 ALDOB 612724 ENO3 131370 G6PC 613742 GAA 606800 GBE1 607839 GYG1 603942 GYS1 138570 GYS2 138571 LAMP2 309060 LDHA 150000 PC 608786 PCK1 614168 PCK2 614095 PFKM 610681 PGAM2 612931 PGM1 171900 PHKA1 311870 PHKA2 300798 PHKB 172490 PHKG2 172471 PRKAG2 602743 PYGL 613741 PYGM 608455 SLC16A1 600682 SLC2A2 138160 SLC37A4 602671	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Glycogen Storage Disease Type III (232400); Glycogen Storage Disease Type IA (232200); Glycogen Storage Disease Type II (232300); Glycogen Storage Disease Type IV (232500); Glycogen Storage Disease Type 0 (240600); Glycogen Storage Disease Type VII (232800); Glycogen Storage Disease Type IXd (300559); Glycogen Storage Disease Type IXa1 (306000); Glycogen Storage Disease Type IX (261750); Glycogen Storage Disease Type IXc (613027); Glycogen Storage Disease Type VI (232700); Glycogen Storage Disease Type V (232600); Glycogen Storage Disease Type Ib	Next Generation Sequencing (NGS) and Sanger sequencing technologies		10-Jul-23
GLYCOGEN STORAGE DISEASE TYPE III VIA THE AGL GENE [9349]	476	AGL (610680)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Glycogen Storage Disease Type III (GSDIII) (232400)	Sanger sequencing		10-Jul-23
GLYCOGEN STORAGE DISEASE TYPE IV VIA THE GBE1 GENE [5431]	476	GBE1 (607893)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Glycogen Storage Disease Type IV (232500)	Sanger sequencing		10-Jul-23
Goose Feather (e70) IgE, Serum	1419		Gold SST	Check if available at CHUM				7-Oct-19

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Granulocyte Antibody Screen, Serum [LAGGN]	1836		Red Top ONLY. SST are NOT acceaptable. Store at 4°C 30 d. Store frozen 1 yr.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Includes neutrophils	Flow cytometry		3-Oct-25
Granulocyte Monocyte - Colony Stimulating Factor, Plasma [GMCSF]	1465		Plasma EDTA. Draw & place on ice. Freeze with 2 h. Stable 21 days frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		bead-based multiplex immunoassay		28-Jan-25
GRHPR Gene, Full Gene Analysis [GRHMS]		GRHPR (604296)	Lavender top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Hyperoxaluria, primary, type II (296000)	Sanger sequencing and. MLPA		
Growth Hormone Releasing Hormone (GH-RH)			3 ml serum (Red or Gold Top) or EDTA plasma should be collected and separated as soon as possible. Freeze the plasma immediately after separation. Minimum specimen size is 1 ml.	InterScience Institute				19-Jan-23
Haloperidol, Serum,/Plasma	115		Red Top or Li heparin (light Green Top)	In-Common Laboratories		LC-MS		9-Sep-21
Hearing Loss: Branchio-Oto-Renal (BOR) Syndrome (115630)		EYA1 (601653)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. EYA1 gene sequencing 2. EYA1 gene dossage	To be verified.	
Hearing Loss: Non-Syndromic (Connexin 26 & 30)		GJB2 (121011); GJB6 (604418)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. GJB2 sequencing and GJB6 deletion 2. GJB2 sequencing only	To be verified.	

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Heparin Induced Thrombocytopenia Testing [HIT]	116		1) RED TOP. 4 mL. Draw blood into red-top vacutainer and allow to clot. Centrifuge and transfer serum to two plastic tube. 2) Freeze promptly. Ship frozen	Platelet Immunology Laboratory, McMaster University		HIT Confirmatory Test (Serotonin Release Assay; SRA) $500; HIT Screen test (Anti-PF4/heparin EIA) $200; SRA & EIA $500		24-Aug-23
Hereditary Angioedema Gene Panel I; Hereditary Angioedema Panel Gene Panel II	1505	Panel I: SERPING1, F12, KNG1, ANGPT1; Panel II: SERPING1, PLG, MYOF, HS3ST6	Lavender Top (EDTA)	VIRANT Diagnostics		Next-Generation sequencing		27-Nov-25
Hereditary Cerebral Small Vessel Disease Panel [53701]	1582	COL4A1, COL4A2, COLGALT1, CTC1, CTSA, FOXC1, GLA, HTRA1, NOTCH3, TREX1	Lavender Top (EDTA)	Invitae/Dynacare		Next-Generation sequencing	Replaces Preven Genetics panel	8-Apr-25
HEREDITARY DIFFUSE GASTRIC CANCER (137215) VIA THE CDH1 GENE [7393]		CDH1 (192090)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)				10-Jul-23
Hereditary Erythrocytosis Mutations [HEMP]	1582	EPOR (133171), VHL (608537), EGLN1(PHD2) (606425), EPAS1 (HIF2A) (603349)	Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Erythrocytosis, familial, 1 (133100); Erythrocytosis, familial, 2 (263400); Erythrocytosis, familial, 3 (609820); Erythrocytosis, familial, 4 (611783)	Sanger sequencing. Panel: EPOR: 133171, EGLN1: 606425, EPAS1: 603349, As reflex VHL: 608537		20-Oct-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
HEREDITARY HEMOCHROMATOSIS PANEL [10243]	1655	FTH1 (134770); FTL (134790); HAMP (606464); HFE (613609); HJV (608374); SLC40A1 (60463); TFR2 (604720)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hemochromatosis Type 5 (615517); Hyperferritinemia Cataract Syndrome (600886); Hemochromatosis Type 4 (606069); Hemochromatosis Type 2 (602390); Hemochromatosis Type 2B (613313); Hemochromatosis Type 3 (604250); Hemochromatosis Type 1 (235200)	Next-Generation sequencing		10-Jul-23
Hereditary Hemorrhagic Telangiectasia (HHT) Panel [10131]	242	EPHB4, GDF2, RASA1ENG (131195), ACVRL1 (601284), SMAD4 (600993)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Telangiectasia, hereditary hemorrhagic, type 1 (187300) (OSLER-RENDU-WEBER DISEASE);Telangiectasia, hereditary hemorrhagic, type 2 (600376); Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome (175050)	Sanger sequencing and MLPA		10-Sep-24
Hereditary Hemorrhagic Telangiectasia and Vascular Malforfmations Panel [02352]	242	ENG (131195), ACVRL1 (ALK1) (601284), SMAD4 (600993) EPHB4 GDF2	Lavender Top (EDTA)	Invitae (www.invitae.com)				23-Jan-24
Hereditary Melanoma and Skin Cancer Panel [ON0501]	243		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		16-Sep-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Hereditary Neuropathy Sequencing & Del/Dup Panel [737]	656	AARS, ATL1, ATP7A, BSCL2, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FAM134B, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GLA, HINT1, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KIF1A, KIF5A, LITAF, LMNA, LRSAM1, MFN2, MPZ, MTMR2, NDRG1, NEFL, NGF, NTRK1, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, REEP1 (C2ORF23), SBF2, SCN9A, SH3TC2, SLC12A6, SLC52A2, SPTLC1, SPTLC2, TFG, TRPV4, TTR, WNK1 (exon 10 only), YARS	Lavender Top (EDTA)	GeneDx		NGS		31-May-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Hereditary Spastic Paraplegia: Autosomal Dominant [HSP-Panel 1]	657	ALDH18A1 ATL1 BSCL2 C10orf2 HSPD1 KIAA0196 KIF5A NIPA1 POLG POLG2 REEP1 SPAST RTN2 SLC33A1 SETX	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		NextGen Sequencing	Price of individual genes: $500 (seq) + 500$ (del/dupl)	21-Aug-19

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Hereditary Spastic Paraplegia: Autosomal Recessive [HSP-Panel 2]	657	ALDH18A1 ALS2 AP4B1 AP4E1 AP4M1 AP4S1 AP5Z1 C10orf2 C12orf65 C19orf12 CYP2U1 CYP7B1 DDHD1 DDHD2 ENTPD1 ERLIN1 ERLIN2 FA2H GBA2 GJC2 KIF1A KIF1C NT5C2 PGAP1 PNPLA6 POLG SACS SPG11 SPG20 SPG21 SPG7 TECPR2	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		NextGen Sequencing	Price of individual genes: $500 (seq) + 500$ (del/dupl)	21-Aug-19

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Hereditary Spastic Paraplegia: Comprehensive Testing [HSP-COMP]	657	ALDH18A1 ATL1 BSCL2 C10orf2 HSPD1 KIAA0196 KIF5A NIPA1 POLG POLG2 REEP1 SPAST RTN2 SLC33A1 SETX ALS2 AP4B1 AP4E1 AP4M1 AP4S1 AP5Z1 C10orf2 C12orf65 C19orf12 CYP2U1 CYP7B1 DDHD1 DDHD2 ENTPD1 ERLIN1 ERLIN2 FA2H	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		NextGen Sequencing	Price of individual genes: $500 (seq) + 500$ (del/dupl)	21-Aug-19

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Hereditary Spastic Paraplegia: Deletion & Duplication Analysis [HSP-DOSAGE]	657		Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		Deletion & Duplication analysis by exon targeted microarray		21-Aug-19
Heterotaxy and Situs Inversus NGS Panel	385	CVR2B, CCDC39, CCDC40, CFC1, DNAAF1, DNAAF2, DNAAF3, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, FOXH1, GDF1, INVS, LEFTY2, NKX2-5, NME8, NODAL, ZIC3	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
Histamine, 24-Hour Urine [FH24U]	117		4 mL urine from a 24-hour collection containing 10 mL 6N HCl; Altenate: No presevative. Specimen Stability: Room temperature: 48 hours, Refrigerated: 14 days, Frozen: 14 days. Patient should refrain from taking allergy causing drugs, antihistamines, oral corticosteroids, and substances which block H2 receptors for at least 24 hours prior to specimen collection.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	carcinoid	Immunoassay		13-Jul-21

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Histamine, Plasma [FHSPL]	117		Draw 3 mL blood in a lavender-top (EDTA) tube(s). Cool immediately on ice. Centrifuge at 1500 rpm for 10 minutes at 4 degrees C. The centrifugation should be performed within 20 minutes of collection. Carefully remove 1 mL of EDTA plasma from the upper part of the tube. Freeze and send frozen in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		EIA		1-May-24
Histoplasma Antibody IgG, IgM, EIA [326]	396		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. CSF: Sterile transport tube.STABILITY Room Temperature: 14 days Refrigerated: 14 days Frozen: 14 days	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22
Histoplasma Antibody, Serum [SHSTO]			Red Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Histoplasmosis		Reserved for Microbiology	20-Oct-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Histoplasma Antigen Quantitative EIA [310}	396		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. Plasma: Collect plasma specimens in an EDTA, heparin or sodium citrate tube. Centrifuge for 15 minutes and pipette plasma into a plastic screw cap vial. Urine/CSF/BAL/Other Body Fluid: Submit urine, CSF, BAL and all other body fluids in a sterile screw cap container. STABILITY Room Temperature: 2 weeks Refrigerated: 2 weeks Frozen: Indefinitely	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22
Histplasma Antibody by Immunodiffusion [321]	396		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. STABILITY Refrigerated: 14 days Frozen: 6 months	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22
Hydroxychloroquine, Serum [HCHQ]	1689		Red Top PREFERRED. Store at -20°C (stable 28 d).	In Common Laboratories		LC-MS/MS		25-Nov-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Hyper IgE Syndrome Panel [10169]	1258	DOCK8 (614443), SPINK5 (605010), STAT3 (102582), TYK2 (176941)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hyper-IgE recurrent infection syndrome {147060); Hyper-IgE recurrent infection syndrome, autosomal recessive (243700); Netherton syndrome (256500); Tyrosine kinase 2 deficiency (611521)	Exon Array CGH, Next-gen Sequencing		10-Jul-23
Hyperglycemia and Hypoglycemia via the GCK Gene [8427]		GCK (138079)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hyperinsulinemic Hypoglycemia, Familial 3 (602485); Maturity-Onset Diabetes Of The Young, Type 2 (125851); Permanent Neonatal Diabetes Mellitus (606176)	Sanger sequencing		10-Jul-23
Hyperglycosylated hCG			Gold SST	Quest Laboratories	First trimester screen for Down syndrome and trisomy 18		To be verified.	

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Hyperlipidemia Panel Plus Analysis [CA1101]	1533	ABCA1 (600046), ABCG5 (605459); ABC8 (603076); APOA1 (107680); APOB (143890); APOC3 (107720); APOE (107741); LDLR (144010); LDLRAP1 (81479); LPL (699708); PCSK9 (603776)	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/	Familial Hypercholesterolemia (143890); Hypercholesterolemia, Autosomal Dominant, Type B (144010); Hypercholesterolemia, Autosomal Dominant, 3 (603776); Hypercholesterolemia, Autosomal Recessive (603813)	Next Generation DNA Sequencing + Del/Dupl	Family Member testing $450. Available at ldC.	16-Jun-17

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Hypoglycemia Panel [12057]	433	ACADM 607008 ACADVL 609575 ACAT1 607809 ACSF3 614245 AGL 610860 ALDOB 612724 CA5A 114761 DGUOK 601465 ETFA 608053 ETFB 130410 ETFDH 231675 FBP1 611570 G6PC 613742 GALT 606999 GK 300474 GYS2 138571 HADH 601609 HMGCL 613898 HMGCS2 600234 MLYCD 606761 MPV17 137960 NNT 607878 OXCT1 601424 PC 608786 PCK1 614168 PCK2 614095 PGM1 171900 PHKA2 300798 PHKB 172490 PHKG2 172471	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency AR 246450 Alpha-Methylacetoacetic Aciduria AR 203750 Combined Malonic And Methylmalonic Aciduria AR 614265 Congenital Disorder of Glycosylation, Type It AR 614921 Fanconi-Bickel Syndrome AR 227810 Fructose-Biphosphatase Deficiency AR 229700 Galactosemia AR 230400 Glycogen Storage Disease 0, Liver AR 240600 Glycogen Storage Disease Type Ia AR 232200 Glycogen Storage Disease Type Ib AR 232220 Glycogen Storage Disease Type Ic AR 232240 Glycogen Storage Disease Type III AR 232400 Glycogen Storage Disease Type IXa1 XL 306000 Glycogen Storage Disease Type IXc AR 613027 Glycogen Storage Disease Type VI AR 232700 Glycogen Storage DiseaseType IXb AR 261750 Hereditary Fructose Intolerance AR 229600 Malonyl-CoA Decarboxylase Deficiency AR 248360 Monocarboxylate Transporter 1 Deficiency AD,AR 616095 Phosphoenolpyruvate Carboxykinase Deficiency, Cytosolic AR 261680 Phosphoenolpyruvate Carboxykinase Deficiency, Mitochondrial AR 261650 Pyruvate Carboxylase Deficiency AR 266150 Succinyl-CoA Acetoacetate Transferase Deficiency AR 245050	NextGen Sequencing		25-Feb-25
Hypomyelinating leukodystrophy 7 and 8 (4H syndrome, 607694 and 614381)	964	POLR3A (614258); POLR3B (614366)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
HYPOMYELINATION AND CONGENITAL CATARACT (HCC) VIA THE HYCC1/FAM126A GENE [11317]		FAM126A (610531)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hypomyelination And Congenital Cataract (610532)	Sanger sequencing		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Hypophosphatasia via the ALPL Gene [851]		ALPL (171760)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Adult Hypophosphatasia (146300); Childhood Hypophosphatasia (241510); Infantile Hypophosphatasia (241500)			11-Jun-18
IA-2 Antibodies, Serum [IA2QST]]	35		Gold SST or Red Top. Specimen Stability - Room temperature: 7 days Refrigerated: 7 days Frozen: 30 days	In-Common Laboraotories	Synonyms:anti-tyrosine phosphatase or anti-Islet antibody; ICA-512	ELISA		25-Feb-25
IBD sgi Diagnostic [1800]	120		2.0 mL Serum (Red Top or SST) AND 2.0 mL Whole Blood EDTA / Lavender Top Tube. Store at 4°C.	Prometheus Biosciences	differentiate among IBD types			
Ibuprofen (Motrin, Advil, Nuprin), serum [FIBUP]			Collect Plain Red. Also acceptable: Green top. NO GEL. Specimen Preparation Separate from cells. Transfer 1 mL serum or plasma to plastic vial. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Optimize drug therapy and monitor patient adherence.	HPLC-UV		20-Nov-17
Ichthyosis Panel [DE0601]	1216	SLC27A4 (604194)	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/	ichthyosis prematurity syndrome (608649}	NGS		10-Jul-23
Idiopathic Ataxia anti MPP-1	1465		Gold SST or Red Top	Mitogen Advanced Diagnostics				13-Jan-26

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Idiopathic Generalized Epilepsy Panel	1031	ADSL, ALDH7A1, ARHGEF9, ARX, ATP6AP2, ATRX, CACNA1A, CACNA1H, CACNB4, CASK, CASR, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CNTN2, CNTNAP2, CPA6, CSTB, CUL4B, DCX, DEPDC5, DHFR, DNAJC5, DYNC1H1, EFHC1, EPM2A, FGD1, FOXG1, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, GPC3, GRIA3, GRIN2A, HSD17B10, KANSL1, KCNC1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, LGI1, MBD5, ME2, MECP2, MEF2C, NHLRC1, NIPA2, NRXN1, OFD1, OPHN1, PAK3, PCDH19, PHF6, PIGA, PLP1, PQBP1, PRICKLE1, PRICKLE2, PRRT2, RAB39B, ROGDI, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC2A1, SLC9A6, SMC1A, SRPX2, STX1B, STXBP1, SYN1, SYNGAP1, SYP, TBC1D24, TCF4, UBE3A, ZEB2	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)				3/26/2025
IGF Binding Protein-1 (IGFBP-1) [FIGBP]			Draw blood in a plain, red-top tube(s). Spin down and separate within one hour. Ship 0.5 mL frozen in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		RIA		20-Nov-17

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
IGF-2 [FFIG2]	123		Draw blood in a RED TOP OR GOLD SST acceptable. Separate within 1 hour of collection. Freeze immediately. STABLE 14 Days at -20°C.	Mayo Clinical Laboratories (Esoterix)				17-Dec-25
IL-2 by Luminex [LIL2]	1523		Plasma from a lavender top (EDTA) tube is the only acceptable sample type. Mix sample thoroughly. Specimen Preparation: Centrifuge at room temp within one half hour of collection; preferably immediately after venipuncture. Transfer the cell-free plasma to a clean tube and immediately freeze the cell-free plasma on dry ice or at -70°C.	National Jewish Health		Luminex		18-Mar-21
IL12Rß1 (CD212) [IL12PATHWAY]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)			To be accompanied by normal control sample	14-Oct-20
Immune Complexes, Serum/Plasma [C1QBIND]	1465		Gold SST. Store at -20°C.	In Common Laboratories				20-Aug-25
Inclusion body myopathy and autosomal recessive, ealrly onset myopathy via the MYH2 gene [9837]		MYH2 (160740)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Inclusion Body Myopathy 3 (605637)	Sanger sequencing	Transfered to HSJ.	10-Jul-23
Infliximab et Ac anti-infliximab [30098]			Serum. Stable 48 h at RT, 5 days at 4°C, or 30 days at -20°C.	L'Hôpital Maisonneuve-Rosement		ELISA		
INGγR12 (CD119) [ILFGPATHWAY]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)			To be accompanied by normal control sample	14-Oct-20
Inhibin A, Tumor Marker, Serum [INHA]			Red Top. Store at 4°C (7 days) or frozen (90 days).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Inhibin B available at CHUM (Gold Top)	Sequential 2-Step Immunoenzymatic Assay	Available at SJH	18-Jul-17
Insulin Antibodies [INSAB]	853		Gold SST or Red Top	In Common Laboratories		RIA		25-Feb-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Insulin Receptor		INSR (147670)	Lavender Top (EDTA) 2-4 cc (1-2 cc for children less than 1year old)	Fulgent Genetics (fulgentdiagnostics.com)	Hyperinsulinemic hypoglycemia familial 5 (609968); Insulin-resistant diabetes mellitus AND acanthosis nigricans (610549); Leprechaunism syndrome (246200); Pineal hyperplasia AND diabetes mellitus syndrome (262190)	NGS and CGH		3/26/2025
Insulin-Like Growth Factor 1 (IGF1), LC-MS and Insulin-Like Growth Factor-Binding Protein 3 (IGFBP3) Growth Panel [IGFGP]	123		Red Top. Split into 2 plastic vials.Store and send serum frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		IGFMS: Liquid Chromatography-Mass Spectrometry (LC-MS) IGFB3: Enzyme-Labeled Chemiluminescent Immunometric Assay	Not M-R	27-Aug-23
Insulin-Like Growth Factor 1 [IGF-1]			Red Top	Royal Victoria Hospital	somatomedin C			
Intact Fibroblast Growth Factor 23 (FGF23), Serum [IFG23]	448		Serum SST. Stable at -20°C for 90 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELSIA	No longer available at CHUM as in province testing.	23-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Intellectual Disability NGS Panel	920	ABCC6, ABCD1, ABCG5, ACAT1, ACOX1, ACSL4, ACY1, ADAR, ADSL, AFF2, AGL, AGT, AGTR2, AHI1, AIFM1, ALDH18A1, ALDH4A1, ALDH5A1, ALG11, ALG12, ALG6, ALX4, AMER1, ANK3, ANKRD11, AP1S1, AP1S2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, AR, ARG1, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARX, ASPM, ASS1, ATL1, ATP10A, ATP13A2, ATP1A2, ATP6AP2, ATP7A, ATRX, AUH, AUTS2, AVP, AVPR1A, AVPR2, BBS9, BCOR, BCS1L, BDNF, BIN1, BRAF, BRIP1, BRWD3, BUB1B, CACNA1C, CACNG2, CAMTA1, CANT1, CASK, CBS, CC2D1A, CC2D2A, CCDC22, CCDC88C, CDH15, CDK16, CDKL5, CDKN1C, CEP290, CEP41, CEP57, CHD7, CHD8, CHRNA4, CLCN4, CLIC2, CLN3, CNKSR2, CNTNAP2, CNTNAP5, COG5, COG7, COL1A2, CP, CPA6, CPS1, CRADD, CRBN, CREBBP, CTC1, CTNNB1, CTSA, CUL4B, CYB5R3, CYP27A1, D2HGDH, DARS2, DBT, DCX, DHCR24, DHCR7, DKC1, DLG3, DLGAP2, DMD, DOCK4, DPP10, DPP6, DPYD, DYNC1H1, DYRK1A, EBP, EFNB1, EHMT1, EIF2S3, ELOVL4, ERCC2, ERCC3, ERCC5, ERCC6, ERCC8, FAAH2, FAM126A, FANCB, FANCG, FBLN5, FBN1, FGD1, FGF14, FGFR1, FGFR2, FGFR3, FKRP, FKTN, FLNA, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FRMPD4, FTO, FTSJ1, G6PC3, GABRB3, GABRG1, GABRG2, GALE, GAMT, GAN, GBA, GBE1, GCK, GDI1, GFAP, GFM1, GHR, GK, GLI3, GLRA1, GLUL, GLYCTK, GM2A, GNA14, GNAS, GNPAT, GNPTAB, GNPTG, GPC3, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRM1, GRPR, GSPT2, GSS, GUSB, GYS2, HAX1, HCCS, HCFC1, HDAC4, HDAC8, HECW2, HEPACAM, HEXB, HOXA1, HOXD10, HPD, HPRT1, HSD17B10, HSPD1, HUWE1, IDS, IGBP1, IGF1, IGF1R, IL1RAPL1, IMMP2L, INSR, IQSEC2, IRX5, ITGA7, KATNAL2, KCNJ10, KCNJ11, KCNK9, KCNQ2, KCTD13, KCTD7, KDM5C,	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing	Available at HSJ.	26-Mar-25
Interferon Beta Neutralizing Antibodies In MS Patient Treated With IFN [P91858]			Gold SST	Neuro-Immunology Laboratories (UBC)				
Interlukin 2 Receptor, Soluble {FIL2S]	927		Serum. Store at -20°C. Stable 1 year.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	hemophagocytic lymphohistiocytosis (HLH)	Liminex Bead assay.	Sent though MCL. Performed at ARUP.	27-Jan-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Interstitial Lung Disease Antibody (ILD) Profile	1724		Gold SST or Red Top. Does NOT accept CSF.	Mitogen Advanced Diagnostics		LIA		13-Jan-26
Iron-refractory Iron Deficiency Anemia (TMPRSS6 Single Gene Test)		TMPRSS6 (609862)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Iron refractory iron deficiency anemia (206200)	NextGen Sequencing		3/26/2025
ITGA9 - Single Gene Testing		ITGA9 (603963)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Integrin alpha 9	NextGen Sequencing		3/26/2025
JAG1 Gene Sequencing & Del/Dup [1004]		JAG1 (601920)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	Alagille syndrome (118450)			15-Sep-22
Jaundice Chip			Lavender Top (EDTA)	Cinncinatti Children's Hospital (Division of Human Genetics Diagnostic Laboratories)	multiple genes	gene chip		

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
JOUBERT AND MECKEL-GRUBER SYNDROMES NEXTGEN SEQUENCING (NGS) PANEL [10387]	332	HI1 (608894) ARL13B (608922) B9D1 (614144) B9D2 (611951) C5orf42 (614571) CC2D2A (612013) CEP290 (610142) CEP41 (610523) INPP5E (613037) KIF7 (611254) MKS1 (609883) NPHP1 (607100) NPHP3 (608002) OFD1 (300170) RPGRIP1L (610937) TCTN1 (609863) TCTN2 (613846) TCTN3 (613847) TMEM138 (614459) TMEM216 (613277) TMEM231 (614949) TMEM237 (614423) TMEM67 (609884) TTC21B (612014) ZNF423 (604557)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NextGen Sequencing		10-Jul-23
Juvenile Polyposis Syndrome (610069 and 174900) [536]		BMPR1A (601299); SMAD4 (600993)	Lavender Top (EDTA)	GeneDx (www.genedx.com)		Tier 1: Sanger Sequencing, Exon Array CGH		15-Sep-22
Juvenile Polyposis Syndrome (610069 and 174900) [537]		BMPR1A (601299)	Lavender Top (EDTA)	GeneDx (www.genedx.com)		Tier 2: sequencing		15-Sep-22
Juvenile Polyposis Syndrome (610069 and 174900) [538]		BMPR1A (601299); SMAD4 (600993)	Lavender Top (EDTA)	GeneDx (www.genedx.com)		Tier 3: Exon Array CGH		15-Sep-22
KCNA1 Full Gene Sequencing Analysis [MOL064]		KCNA1 (176260)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Episodic ataxia/myokymia syndrome			9-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Kidney Dysplasia NGS Panel	1615	ACE, AGT, AGTR1, ANOS1, BMP4, BMP7, CDC5L, CHD1L, DSTYK, EYA1, FGF20, FGFR2, FRAS1, FREM1, FREM2, GATA3, GLI3, GRIP1, HNF1B, HOXA13, HOXA4, HOXB6, HPSE2, ITGA8, LRP4, MUC1, MYH9, NIPBL, PAX2, REN, RET, ROBO2, SALL1, SIX1, SIX2, SIX5, SOX17, TRAP1, UMOD, UPK3A, WNT4	Lavender Top (EDTA) [2 tubes]	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
KIT Single Gene	1537	cKit (164920)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Systemic mastocytosis (154800)	NextGen Sequencing		3/26/2025
KRABBE DISEASE VIA THE GALC GENE [7883]		GALC (606890)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Galactosylceramide Beta-Galactosidase Deficiency (245200)	NGS	enzyme activity at HSJ	10-Jul-23
L1CAM (fetal sexing)		L1CAM (308840)	Gold SST	Laboratoire Cerba (www.lab-cerba.com)				
L1CAM Gene Sequencing & Del/Dup [552]		L1CAM (308840)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	X-Linked Hydrocephalus Syndrome (307000); Spastic Paraplegia 1 (303350); Corpus Callosum, Partial Agenesis Of, X-Linked (304100)	1. Sanger sequencing and aCGH 2. targeted mutation		15-Sep-22
Lacosamide, Serum [LACO]	1572		RED OR GOLD TOP. Store frozen	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		HPLC-MS		28-Jun-18

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
LAMINOPATHIES VIA THE LMNA GENE [347]		LMNA (150330)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Charcot-Marie-Tooth Disease Type 2B1 (605588); Limb-Girdle Muscular Dystrophy, Type 1B (159001); Lipodystrophy, Familial Partial, Dunnigan Type (151660); Dilated Cardiomyopathy 1A (115200); Emery-Dreifuss Muscular Dystrophy, Autosomal Dominan (151350)t; Restrictive Dermopathy, Lethal (275210); Hutchinson-Gilford Syndrome (176670)	Sanger sequencing	Transfered to HSJ.	8-Nov-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Leigh disease and Leigh-like syndromes NextGen DNA Sequencing Panel (75 genes) [NGS351]		AIFM1 (300169); ALDH5A1 (610045); ARX (300382); BCS1L (603647); C12orf65 (613541); COA5 (613920); COX10 (602125); COX14 (614478); COX15 (603646); COX6B1 (124089); CPT2 (600650); DLAT (608770); DLD (238331); ETHE1 (608451); FARS2 (611592); FASTKD2 (612322); FOXRED1 (613622); GCDH (608801); KCNQ2 (602235); LIAS (607031); LRPPRC (607544); MTFMT (611766); MUT (609058); NDUFA1 (300078); NDUFA10 (603835); NDUFA11 (612638); NDUFA12 (614530); NDUFA2 (602137); NDUFA9 (603834); NDUFAF1 (606934); NDUFAF2 (609653); NDUFAF3 (612911); NDUFAF4	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Leigh syndrome (256000); Leigh syndrome, X-linked (308930)	NextGen Sequencing		9-May-20
Leptin [FLEP]	1465		Draw blood in a plain, red-top tube(s). (Serum gel tube is acceptable.) Separate and freeze within one hour. Send 1 mL of serum frozen in a plastic vial. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELISA		9-Oct-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Leukemia, Philadelphia chromosome-positive, resistant to imatinib		ABL1 (189980)	Lavender Top (EDTA)	University Health Network (Toronto General Hospital)				
Leukemia/Lymphoma Immunophenotyping (PNH and ZAP-70 available) [2001]	1465		Na heparin tube. Store and send at RT. Send immediately overnight.	Hematologics, Inc. 3161 Elliot Ave. Suite 200, Seattle WA 98121 1800-860-0934		Flow cytometry		23-Jun-20
Leukotriene E4, 24 Hour, Urine [TLTE4]	1465		Preferred: 24-hour urine collection Container/Tube: Plastic, 5-mL tube (T465) Specimen Volume: 5 mL Collection Instructions: 1. Collect urine for 24 hours.No preservative. 2. Refrigerate specimen during collection, aliquot 5 mL of urine into plastic tube, and send specimen refrigerated. Store frozen (30 days).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS	Replaced by Mast Cell Mediators. Random test available RLTE4	23-Jul-24
Levetiracetam			Lavender Top; transport on ice	Hopital Ste. Justine				
LGI1 and Caspr 2 (VGKC antibodies)	54	LGI1 and Caspr2 (Voltage gated potassium chanel)	1st choice CSF; 2nd choice SERUM has reduced sensitivity	BC Neuroimmunology (bcneuro.ca)				18-Apr-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
LIMB GIRDLE MUSCULAR DYSTROPHY PANEL [10401]	911	ANO5 (608662); CAPN3 (114240); CAV3 (6012530; DES (125660); DNAJB6 (611332); DYSF (603009); FKRP (606596); GMPPB (615320); ISPD (614631); LIMS2 (607908); LMNA (150330); MYOT (604103); PNPLA2 (609059); SGCA (600119); SGCB (600900); SGCD (601411); SGCG (608896); SMCDH1 (614982); TCAP (604488); TNOP3 (610032); TOR1AIP1 (614512); TRAPPC11 (614138); TRIM32 (602290); TTN (188840)	Lavender Top (EDTA) [2 tubes]	Prevention Genetics (www.preventiongenetics.com)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 (607155); Limb-Girdle Muscular Dystrophy, Type 2H (254110);Limb-Girdle Muscular Dystrophy, Type 2G (601954);Limb-Girdle Muscular Dystrophy, Type 2C (253700); Limb-Girdle Muscular Dystrophy, Type 2F (601287); Limb-Girdle Muscular Dystrophy, Type 2E (604286); Limb-Girdle Muscular Dystrophy, Type 2D (608099); Limb-Girdle Muscular Dystrophy, Type 2B (253601); Limb-Girdle Muscular Dystrophy, Type 2A (253600); Limb-Girdle Muscular Dystrophy, Type 2J (608807) ; Limb-Girdle Muscular	NextGen Sequencing (24 genes)	Transfered to HSJ	10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
LIMB GIRDLE MUSCULAR DYSTROPHY TYPE 2B AND MIYOSHI MYOPATHY VIA THE DYSF GENE [3123]	606	DYSF (603009)	Lavender Top (EDTA) [2 tubes]	Prevention Genetics (www.preventiongenetics.com)	Limb-Girdle Muscular Dystrophy, Type 2B (253601); Miyoshi Myopathy (254130), Myopathy, Distal, With Anterior Tibial Onset (606768)		Transfered to MUHC.	8-Nov-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Lipid Metabolism Deficiency NGS Sequencing Panel (71 genes) + Del/Dup + mtDNA [NGS303]		ABCD1 (300371); ABCD2 (601081); ACAA1 (604054); ACAA2 (604770); ACACA (200350); ACACB (601557); ACAD10 (611181); ACAD11 (614288); ACAD9 (611103); ACADL (609576); ACADM (607008); ACADS (606885); ACADSB (600301); ACADVL (609575); ACAT1 (607809); ACAT2 (100678); ACLY (108728); ACOT1 (614313); ACOT12 (614315); ACOT2 (609972); ACOT4 (614314); ACOT6 (614267); ACOT7 (602587); ACOT8 (608123); ACOT9 (300862); ACOX1 (609751); ACOX2 (601641); ACOX3 (603402); ACP6 (611471); ACSBG1 (614362); ACSBG2 (614363); ACSF2 (610465); ACSF3	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		9-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Lipodystrophy		AGPAT2 (603100), AKT2 (164731), BSCL2 (606158), CAV1 (601047), CIDEC (612210) , LMNA (150330), PPARG (601487), PTRF (603198), TBC1D4 (612465), ZMPSTE24 (606480)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
Liquid Biopsy ctDNA Targeted NGS Panel and Low Pass Whole Genome Sequencing (LP-WGS)	1395		STRECK Cell-free DNA BCT tubes ONLY. Ship Mon-Wed only. Store at RT. Stable for 7 days.	Clinical Molecular Diagnostics. Hosital for Sick Children.	Single mutation not performed.	NextGen Sequencing	RESTRICTED. Requires approval by Head of Send Out	1/20/2026
Lissencephaly NGS Panel		ACTB, ACTG1, ARX, DCX, FKRP, FKTN, LARGE, PAFAH1B1, POMGNT1, POMT1, POMT2, RELN, TUBA1A, VLDLR	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)				3/26/2025
Long QT/Brugada Syndrome NGS Panel	285	AKAP9 (604001), ANK2 (106410), CACNA1C (114205), CAV3 (601253), KCNE1 (176261), KCNE2 (603796), KCNH2 (152427), KCNJ2 (600681), KCNJ5 (600734), KCNQ1 (607542), SCN4B (608256), SCN5A (600163), SNTA1 (601017)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Long QT Syndrome (192500)	NextGen Sequencing	Performed at IdC	26-Mar-25
LRBA	1248		Na heparin tube. Store and send at RT. Send immediately overnight.	Alberta Precision Laboratories (Flow Cytometry - Calgary)		Flow cytometry		1-Jun-21

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
LRP4 Autoantibody Test	18		Serum (Gold SST or Red Top). Store at -20°C	BC Neuroimmunology (bcneuro.ca)	For AcR and MUSK negative subjects	IFA		18-Apr-24
Lurasidone [2543SP]	1688		Red Top or Lavender Top only. Store and send serum/plasma frozen.	NMS Laboratories		LC-MS/MS		17-Mar-22
LYMPHEDEMA-DISTICHIASIS SYNDROME VIA THE FOXC2 GENE [8941]		FOXC2 [602402]	Lavender top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Lymphedema-Distichiasis Syndrome (153400)	NGS		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Lysosomal Disease (NextGen Sequencing Panel and Copy Number Analysis; 72 Genes) [NGS313]	474	ADAMTSL2 (612277), AGA (613228), ANTXR2 (608041), ARSA (607574), ARSB (611542), ASAH1 (613468), ATP13A2 (610513), ATP7A (300011), ATP7B (606882), CERS1 (606919), CLN3 (607042), CLN5 (608102), CLN6 (606725), CLN8 (607837), COL11A2 (120290), COL2A1 (120140), CTNS (606272), CTSA (613111), CTSC (602365), CTSD (116840), CTSF (603539), CTSK (601105), DHCR7 (602858), DNAJC5 (611203), DYM (607461), FUCA1 (612280), GAA (606800), GALC (606890), GALNS (612222), GBA (606463), GLA (300644), GLB1 (611458), GM2A (613109), GNE (603824)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next Generation DNA Sequencing		9-May-20
Lysozyme (Muramidase), Plasma [MURA]	1465		Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Spectrophotometric		15-Jan-24
Macroprolactin			Gold SST. Centrifuge and store serum at 4°C.	Verdun Hosipital		1X per week		
Malignant Hyperthermia Suscpetibility Panel [03285]	632	CACNA1S (114208), RYR1 (180901), STAC3		Invitae (www.invitae.com)		NextGen Sequencing		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Mannose-Binding Lectin Deficiency NGS Test		MBL2 (154545)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Mannose-binding protein deficiency (614372)	Next Generation DNA Sequencing		3/26/2025
MARFAN SYNDROME AND RELATED AORTOPATHIES PANEL [1212]	279		Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NextGen Sequencing		10/Jul/23
Marfan Syndrome and Thoracic Aortic Aneurysm and Dissection NGS Panel	279	ACTA2, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, FLNA, MED12, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2 (22 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Marfan syndrome, type I (154700)	NextGen Sequencing		17-Feb-19
MARINESCO-SJOGREN SYNDROME VIA THE SIL1 GENE [11667]		SIL1 (608005)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Marinesco-Sjogren Syndrome (248800)	NGS		10/Jul/23
Mast Cell Mediators, 24 Hour, Urine [MCM24]	1465		24h urine collection. No preservatives. Stable for 28d at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS & enzymatic assay	Replaces separate assays for N-methylhistamine dinor-prostaglandin F2a, & leukotriene E4.	28/Nov/24
Meckel-Gruber syndrome Sequencing Panel		CC2D2A (612013); CEP290 (610142); MKS (609883); RPGRIP1L (610937); TCTN (613846)2; TMEM67 (609884); TMEM216 (613877)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories (dnatesting.uchicago.edu)	Meckel Syndrome, Type 8 (613885); Meckel Syndrome, Type 6 (312284); Meckel Syndrome, Type1 (249000); Meckel Syndrome, Type 4 (611134); Meckel Syndrome, Type 3 (607361); Meckel Syndrome, Type 5 (611561); Meckel syndrome 2 (603194)	Sanger sequencing		

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
MECP2 Analysis		MECP2 (300005)	Lavender Top (EDTA) 2-5 cc	Alberta Children's hospital http://www.medicalgenetics.ca/molecular.html	Rett's disorder (312750)	Sanger sequencing		4-May-17
Medullary Cystic Kidney Disease type 2 and Familial Juvenile Hyperuricemic Nephropathy type 1 via the UMOD Gene [8209]		UMOD (191845)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Familial Juvenile Hyperuricemic Nephropathy (162000); Medullary Cystic Kidney Disease 2(603860)	NGS		10-Jul-23
Metformin, Plasma [FMETN]	1982		Lavender Top only. Store at 4°C. Stable 30 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		HPLC-TMS		18-Apr-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Microcephaly NGS Panel	350	AKT3, ANKLE2, ARFGEF2, ASPM, ATR, ATRIP, BUB1B, CASK, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CEP135, CEP152, CEP63, CKAP2L, COX7B, CRIPT, DIAPH1, DNM1L, EFTUD2, HMGB3, IER3IP1, KATNB1, KIF11, KNL1, LIG4, MCPH1, MED17, MFSD2A, MIR17HG, MRE11, MSMO1, MYCN, NBN, NDE1, NHEJ1, NIN, NR2E1, PAFAH1B1, PCLO, PCNT, PHC1, PLEKHG2, PLK4, PNKP, POMT1, PPP1R15B, PQBP1, QARS, RARS2, RBBP8, RTTN, SASS6, SLC1A4, SLC25A19, SLC9A6, SPATA5, STAMBP, STIL, THOC6, TRMT10A, TSEN2, TSEN34, TSEN54, TUBB2B, TUBGCP4, TUBGCP6, VRK1, WDR62, WDR73	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
Migraine Panel [NE1201]	581		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		Next-Generation sequencing		21-Aug-20
Minimal Residual Disease Testing (MRD)	1084		Na heparin tube. Store and send at RT. Send immediately overnight.	Toronto General Hospital		Flow cytometry	Prices can vary	13-Nov-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Minimal Residual Disease Testing (MRD) PreB-ALL	1084		Na heparin tube. Store and send at RT. Send immediately overnight.	Johns Hopkins Medical Laboratories		Flow cytometry	Price can vary by number of markers; 9-15 markers quoted here	28-Nov-23
Minimal Residual Disease Testing (MRD) T cell [2002]	1084		Na heparin tube. Store and send at RT. Send immediately overnight.	Hematologics, Inc. 3161 Elliot Ave. Suite 200, Seattle WA 98121 1800-860-0934		Flow cytometry		10-Jul-23
Minimal Residual Disease Testing (MRD) T-ALL	1084		Na heparin tube. Store and send at RT. Send immediately overnight.	Johns Hopkins Medical Laboratories		Flow cytometry	Price can vary by number of markers; 9-15 markers quoted here	14-Nov-23
Minimal Residual Disease Testing for Acute Lymphoblastic Leukemia	1084		EDTA Whole Blood. Keeep at room temperature.	Flow Cytometry Laboratory. London Health Sciences Centre.	For Friday samples email Ben Hedley with ID information & location where sample is being sent.	Flow cytometry		4-Aug-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Mitochondrial Genome Sequencing [MOL021]	518	The human mitochondrial genome is 16569 base pairs in length and encodes 37 genes including 2 ribosomal RNA genes, 22 transfer RNA genes, and 13 protein-coding genes. Our mitochondrial DNA (mtDNA) panel includes complete sequencing of the following genes (MIM#): MTRNR1 (561000), MTRNR2 (561010); MTTA (590000); MTTR (590005); MTTN (590010); MTTD (590015); MTTC (590020); MTTE (590025); MTTQ (590030); MTTG (590035); MTTH (590040); MTTI (590045); MTTL1 (590050); MTTL2 (590055); MTTK (590060); MTTM (590065); MTTF (590070); MTTP (590075); MTTS1 (590080); MTTS2 (590085); MTTT (590090); MTTW (590095); MTTY (590100); MTTV (590105); MTND1 (516000); MTND2 (516001); MTND3 (516002); MTND4 (516003); MTND4L (516004); MTND5 (516005); MTND6 (516006); MYCYB	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Mitochondrial DNA Deletion Syndromes Mitochondrial DNA Depletion Syndrome Mitochondrial DNA Depletion Syndrome Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form Mitochondrial DNA Depletion Syndrome, Hepatocerebral Form Mitochondrial DNA Depletion Syndrome, MNGIE Form Mitochondrial DNA Depletion Syndrome, Myopathic Form mtDNA Deletion Syndromes	NextGen Sequencing	Available at HSJ	9-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Mitochondrial Genome Sequencing + Deletion Analysis [MOL189]	518	MTRNR1 (561000), MTRNR2 (561010); MTTA (590000); MTTR (590005); MTTN (590010); MTTD (590015); MTTC (590020); MTTE (590025); MTTQ (590030); MTTG (590035); MTTH (590040); MTTI (590045); MTTL1 (590050); MTTL2 (590055); MTTK (590060); MTTM (590065); MTTF (590070); MTTP (590075); MTTS1 (590080); MTTS2 (590085); MTTT (590090); MTTW (590095); MTTY (590100); MTTV (590105); MTND1 (516000); MTND2 (516001); MTND3 (516002); MTND4 (516003); MTND4L (516004); MTND5 (516005); MTND6 (516006); MYCYB (516020); MTCO1	Blood | Fibroblasts | Muscle | Extracted DNA | Buccal Cells	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Leber hereditary optic neuropathy (LHON); Neuropath, ataxia, and retinitis pigmentosa (NARP); Myoclonic epilepsy associated with ragged-red fibers (MERRF); Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)	Deletion; Capillary DNA Sequencing	Availabe at HSJ	14-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Mitochondrial Genome Sequencing + Deletion Analysis + Depletion Testing (Leukocyte) [MOL232]	518	The human mitochondrial genome is 16569 base pairs in length and encodes 37 genes including 2 ribosomal RNA genes, 22 transfer RNA genes, and 13 protein-coding genes. Our mitochondrial DNA (mtDNA) panel includes complete sequencing of the following genes (MIM#): MTRNR1 (561000), MTRNR2 (561010); MTTA (590000); MTTR (590005); MTTN (590010); MTTD (590015); MTTC (590020); MTTE (590025); MTTQ (590030); MTTG (590035); MTTH (590040); MTTI (590045); MTTL1 (590050); MTTL2 (590055); MTTK (590060); MTTM (590065); MTTF (590070); MTTP (590075); MTTS1 (590080); MTTS2 (590085); MTTT (590090); MTTW (590095); MTTY (590100); MTTV (590105); MTND1 (516000); MTND2 (516001); MTND3 (516002); MTND4 (516003); MTND4L (516004); MTND5 (516005); MTND6 (516006); MYCYB	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Mitochondrial diseases including: Leber hereditary optic neuropathy (LHON); Neuropath, ataxia, and retinitis pigmentosa (NARP); Myoclonic epilepsy associated with ragged-red fibers (MERRF); Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)	Deletion; Real-time Quantitative PCR analysis; Sequencing	Available at HSJ	14-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Mitochondrial Genome Sequencing + Deletion Analysis + Depletion Testing (Muscle) [MOL340]	518	The human mitochondrial genome is 16569 base pairs in length and encodes 37 genes including 2 ribosomal RNA genes, 22 transfer RNA genes, and 13 protein-coding genes. Our mitochondrial DNA (mtDNA) panel includes complete sequencing of the following genes (MIM#): MTRNR1 (561000), MTRNR2 (561010); MTTA (590000); MTTR (590005); MTTN (590010); MTTD (590015); MTTC (590020); MTTE (590025); MTTQ (590030); MTTG (590035); MTTH (590040); MTTI (590045); MTTL1 (590050); MTTL2 (590055); MTTK (590060); MTTM (590065); MTTF (590070); MTTP (590075); MTTS1 (590080); MTTS2 (590085); MTTT (590090); MTTW (590095); MTTY (590100); MTTV (590105); MTND1 (516000); MTND2 (516001); MTND3 (516002); MTND4 (516003); MTND4L (516004); MTND5 (516005); MTND6 (516006); MYCYB	50-75 milligrams muscle snap frozen in liquid nitrogen and maintained at -80°Celsius or below. (2) DNA extracted from muscle	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Mitochondrial diseases including: Leber hereditary optic neuropathy (LHON) Neuropath, ataxia, and retinitis pigmentosa (NARP) Myoclonic epilepsy associated with ragged-red fibers (MERRF) Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)	Polymerase Chain Reaction (PCR) followed by DNA sequencing analysis; Polymerase Chain Reaction (PCR) followed by Restriction Endonuclease digestion and gel separation; Real-time Quantitative PCR analysis	Available at HSJ	14-May-20
Mitochondrial Respiratory Chain Enzyme Analysis (ETC)	1269		Skin Fibroblasts [3210]; Skeletal Muscle Biopsy [3200]; 150 mg; Store at -80°C.	Baylor Genetics. 2450 HOLCOMBE BLVD. GRAND BLVD. RECEIVING DOCK HOUSTON, TX 77021-2024		Spectrophotometric		3-Oct-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
MLPA (screen for deletions of CFHR1-CFHR3)		CFHR1 (134371); CFHR3 (605336)	Lavender Top (EDTA)	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	Hemolytic uremic syndrome, atypical, susceptibility to (235400)	MLPA		1-Mar-25
MODY Neonatal Diabetes NGS Panel	481	ABCC8, AKT2, BLK, CEL, CISD2, CP, EIF2AK3, FOXP3, GATA6, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, IER3IP1, INS, INSR, KCNJ11, KLF11, NEUROD1, NEUROG3, PAX4, PDX1, PTF1A, RFX6, SLC2A2, WFS1, ZFP57 (30 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
Mold Mix 1	1465		Serum (Gold SST or Red Top)	In-Common Laboratories				12-Mar-20
Mold Mix 2	1465		Serum (Gold SST or Red Top)	In-Common Laboratories				12-Mar-20
Molecular Testing for Lissencephaly		DCX (300121)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories (dnatesting.uchicago.edu)	Lissencephaly, X-linked (300067);Subcortical laminal heteropia, X-link (300067); testing for a known mutation	Sanger sequencing		
Monocyte Type I and Type II Interferon (IFN) Signature Quantitation by Flow Cytometry [T1A2MP]	1465		Whole Blood (EDTA or Na heparin). Keep at RT, Stable for 72 h at RT.	Nationwide Children's Hospital. Laboratory Services Room C1955. Diagnostic immunology Lab, Colombus, OH. 1800-934-6575			Send Out must be advised the day before sending the sample. Not accepted after 3 pm Thursday.	6-Feb-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Monogenic Autoimmunity Panel [08150]	817	ACP5 ADA2 ADAR AICDA AIRE AP3B1 BLOC1S6 BTKCASP10 CASP8 CD27 CD40LG CR2 CTLA4 CYBA CYBBDOCK8 FADD FAS FASLG FOXP3 ICOS IFIH1 IL10IL10RA IL10RB IL21 IL21R IL2RA ITCH ITK LRBALYST MAGT1 NCF2 NCF4 NFAT5 NFKB2 NFKBIA ORAI1PIK3CD PIK3R1 PLCG2 PNPPRF1 PRKCD RAB27A RAC2RFX5 RFXANK RFXAP RMRP RNASEH2A RNASEH2B RNASEH2C SAMHD1SH2D1A SLC7A7 STAT1 STAT3 STAT5B STIM1 STX11 STXBP2TBX1 TMEM173 TNFRSF13B TNFRSF13C TNFSF12 TPP2 TREX1 UNC13DUNG WAS XIAP	Lavender Top (EDTA) [2 tubes]	Invitae (www.invitae.com)		NextGen Sequencing		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Monogenic Inflammatory Bowel Disease Panel (Invitae) [08122]	1151	ADA, ADAM17, AICDA, BTK, CD3G, CD40LG, CTLA4, CYBA, CYBB, DCLRE1C, DKC1, DOCK8, FOXP3, G6PC3, ICOS, IL10, IL10RA, IL10RB, IL21, IL2RA, IL2RG, ITGB2, LIG4, LRBA, MEFV, MVK, NCF2, NCF4, NFAT5, NLRC4, PIK3CD, PIK3R1, PLCG2, RAG1, RAG2, RTEL1, SH2D1A, SLC37A4, STAT1, STAT3, STIM1, STXBP2, TTC7A, WAS, XIAP, ZAP70	Lavender Top (EDTA)	Invitae (www.invitae.com)	XIAP X-linked lymphoproliferative syndrome 2; ZAP70 Severe combined immunodeficiency, Omenn syndrome; WAS Wiskott–Aldrich syndrome; TTC7A Gastrointestinal defects and immunodeficiency (GIDID) syndrome; STXBP2 Familial hemophagocytic lymphohistiocytosis type 5; STIM1 STIM1 deficiency; STAT3 IPEX-like syndrome; STAT1 IPEX-like syndrome; SLC37A4 Glycogen storage disease type Ib; SH2D1A X-linked lymphoproliferative syndrome 1; RTEL1 Dyskeratosis congenita; RAG2 Severe combined immunodeficiency, Omenn syndrome; RAG1 Severe combined immunodeficiency, Omenn syndrome; PLCG2 Familial cold autoinflammatory	Next-Generation sequencing		10-Jul-23
Monogenic Obesity Panel [KI1701]	485		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		Next Generation DNA Sequencing		19-Aug-25
MTHFR 677C>T	9999							2-Dec-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Muscle Specific Tyrosine Kinase Antibodies (MuSK Ab) [P91022]	43		Gold SST. SERUM ONLY. Centrifuge and transfer serum to plastic container. Store at -20°C. Transport frozen.	BC Neuroimmunology (bcneuro.ca)	myasthenia gravis	SPR	NOTE: test sensitivity is ~100% but has low specificity.. Orderable only by Neurology due to low specificity as SECOND LINE test.	18-Apr-24
Myelin Basic Protein [663]	1465		CSF. Store frozen. Specimen Stability : Room temperature: 7 days. Refrigerated: 14 days. Frozen: 21 days	Quest Laboratories	The pesence of myelin basic protein in the spinal fluid is supportive evidence for the diagnosis of multiple sclerosis and other demyelinating diseases, although it is a non-specific finding and present in other causes of damage to CNS myelin.	RIA		23-Jul-24
Myelin-Associated Glycoprotein (MAG) IgM Antibodies, Serum [AMAGS]	40		Gold SST. Store at -20°C.	In-Common Laboratories		ELSIA	Replaces Mitogen MAG testing	9-Mar-23
Myeloid NGS Screen	1395		Lavender Top (EDTA)	Molecular Genetics Laboratory - LHSC		Next Generation DNA Sequencing	$890 full panel; ≤2 gene fusions $350 each if ordered separately. Transfered to HMR.	28-Oct-21
Myopathy-Rhabdomyolysis		ACAD9, ACADL, ACADM, ACADVL, AGL, C10ORF2, CPT1B, CPT2, GAA, GYS1, HADHA, HADHB, OPA1, OPA3, PFKM, PGAM2, PGM1, PHKA1, POLG, POLG2, RRM2B, SUCLA2, TK2, TYMP	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing	Trasnfer to HSJ after Nov 30.	26-Mar-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Myotonia Congenita via the CLCN1 Gene [11179]	607	CLCN1 (118425)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Congenital Myotonia, Autosomal Dominant Form (160800), Myotonia Congenita Autosomal Recessive (255700)	NGS		10-Jul-23
Myotonic Dystrophy 2	607	CNBP (ZNF9) (116955)	Lavender Top (EDTA)	Molecular Genetics Laboratory - BC Children's Hospital & BC Women's Hospital		Repeat-primed PCR (QP-PCR)		17-Dec-25
Myotonic Dystrophy Type I (160900)	607	DMPK (605337)	Lavender Top (EDTA)	Childern's Hospital of Eastern Ontario			Available at CHUQ: Dystrophie myotonique type 1 (Steinert) > MNG	9-Jan-17
N-Methylhistamine, 24 Hour, Urine [NMH24]	117		Collection Instructions: Collect 24-hour urine with no preservatives. Total volume required. Record on both the specimen container and the request form. Urine Preservative Collection Options: Note: The addition of preservative or application of temperature controls must occur within 4 hours of completion of the collection: 6N HCl, 50% Acetic Acid, Na(2)CO(3), Toluene, 6N HNO(3), Boric Acid, Thymol. Store frozen. **Stable 14 days.**	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	mastocytosis	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)	Replaced by Mast Cell Mediators	23-Jul-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Neopterin/**Tetrahydrobiopterin** (CSF) [NC03]	1224	BH4, Neopterine	Collection tube requirement : Call laboratory to obtain appropriate sample collection containers. Each sample collection set consists of 5 numbered centrifuge tubes in a small plastic bag. Tube #3 contains antioxidants necessary to protect the sample from oxidation. One set of tubes is required per patient. The total CSF volume required is 3.5 milliliters. Collection of sample : CSF should be collected directly from the tap needle: Collect from the first drop in to the containers in numerical order. Fill each tube to the marked line (0.5 milliliters in tubes 1, 2, & 5 – 1.0 ml in tubes 3 & 4). Attach patient identifiers to each tube without covering the tube number. If there is no blood contamination, place the tubes into a biohazard bag and place on ice (or dry ice if available) at the bedside. Transfer the samples to a -80°C freezer ASAP. Ship on dry ice. If the sample is blood contaminated, the tubes should immediately	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Dystonia, dopamine responsive | DRD | GCH1 | Dystonia, dopa responsive, due to sepiapterin reductase deficiency | SPR | Hyperphenylalaninemia, BH4 deficient, B | HPABH4 | Hyperphenylalaninemia, BH4 deficient, A | HPABHA | PTS | Hyperphenylalaninemia, BH4 deficient, C | QDPR | HPABH4C | Hyperphenylalaninemia, BH4 deficient, D | HPABH4D | PCBD1			9-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Nephrotic Syndrome (NS)/Focal Segmental Glomerulosclerosis (FSGS) Panel [10417]	100	ACTN4 604638 ANLN 616027 ARHGAP24 610586 ARHGDIA 601925 CD2AP 604241 COL4A3 120070 COL4A4 120131 COL4A5 303630 COL4A6 303631 COQ2 609825 COQ6 614647 COQ8B 615567 CRB2 609720 CUBN 602997 DGKE 601440 EMP2 602334 FAT1 600976 INF2 610982 ITGA3 605025 ITGB4 147557 KANK1 607704 KANK2 614610 KANK4 614612 LAGE3 300060 LAMA5 601033 LAMB2 150325 LMX1B 602575 MAGI2 606382 MYO1E 601479 NPHS1 602716 NPHS2 604766 NUP107 607617	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Alport Syndrome, Autosomal Dominant AD 104200 Alport Syndrome, Autosomal Recessive AR 203780 Alport Syndrome, X-Linked Recessive XL 301050 Cerebral Palsy, Spastic Quadriplegic, 2 AR 612900 Coenzyme Q10 Deficiency AR 607426 Coenzyme Q10 deficiency, primary, 3 AR 614652 Coenzyme Q10 deficiency, primary, 6 AR 614650 Deafness, X-linked 6 XL 300914 Epidermolysis Bullosa With Pyloric Atresia AR 226730 Epilepsy, Progressive Myoclonic 4, With	Next Generation DNA Sequencing		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Neurofibromatosis (NextGen Sequencing Panel and Copy Number Analysis; 21 Genes) [NGS335]	776	ATM (607585), BRAF (164757), CBL (165360), HRAS (190020), KRAS (190070), MAP2K1 (176872), NF1 (613113), NF2 (607379), NRAS (164790), PTEN (601728), PTPN11 (176876), RAF1 (164760), RIT1 (609591), SDHAF2 (613019), SDHB (185470), SDHC (602413), SDHD (602690), SH3BP2 (602104), SHOC2 (602775), SOS1 (182530), SPRED1 (609291).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next Generation DNA Sequencing		9-May-20
Neurofilament (pNf-H)	1465		CSF	Washington University Neuromuscular Laboratory		ELISA		2-Oct-23
Neurofilament Light Chain (NfL)	1465		EDTA plasma	BC Neuroimmunology (bcneuro.ca)		Lumipulse	Replaces In Common Laboratories	5-Jan-26
Neurofilament Light Chain (NfL), Serum/Plasma [NFLRO S]	1465		Serum, SST or Red Top or Na heparin. Stable at -20°C.	In Common Laboratories (EORLA)	EORLA requisition	Roche Elecsys	Replaced by BC Neuroimmunology	20-Oct-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Neutropenia NGS Panel	836	AP3B1, CSF3R, CXCR4, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1, LAMTOR2, LYST, RAB27A, RAC2, SBDS, SLC37A4, TAZ, USB1, VPS13B, VPS45, WAS, WIPF1	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Cyclical neutropenia (162800), Severe congenital neutropenia autosomal dominant (202700), Severe congenital neutropenia X-linked (300299), Severe congenital neutropenia 4, autosomal recessive (612541), Severe congenital neutropenia 2, autosomal dominant (613107), Severe congenital neutropenia 3, autosomal recessive (610738)	NextGen Sequencing		3/26/2025
Neutrophil Antibody, Flow Cytometry [1606]	1572		Serum. Red Top (only). Room temperature: 7 days Refrigerated: 14 days Frozen: 30 days	Quest Diagnostics				6-Jul-20
NF-1-NG	776	NF-1	Lavender Top (EDTA)	UAB Medical Genomics Laboratory (www.uab.edu/medicine/genetics/medical-genomics-laboratory)		NGS and del/dupl		10-Jul-23
Niemann-Pick Disease Type C Sequencing Panel [3425]	468	NPC1 (607623); NPC2 (601015)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Niemann-Pick Disease Type C1 (257220)	Next Generation DNA Sequencing		Juy 10, 2023
Noonan Syndrome (NextGen Sequencing Panel and Copy Number Analysis; 10 Genes) [NGS414]	778	BRAF KRAS LZTR1 MAP2K1 NRAS PTPN11 RAF1 RIT1 SOS1 SOS2	Lavender top (EDTA) [2 tubes]	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing	Transfer to HSJ on Nov 30	9-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
NOTCH3-CADASIL (125310)	65	NOTCH3 (600276)	Lavender Top (EDTA)	London Laboratories Service Group		Sanger sequencing; 33 coding exons of NOTCH3+ 20nt intronic sequence around each exon	To change to NGS with exon copy number in Oct 2015	
NPM1 MRD	1395	NPM1 (164040)	Lavender Top (EDTA). Bone marrow. Stable 24-48 h at RT after draw. Do not freeze.	Dept of Clinical Laboratory Genetics. Genome Diagnostics. Toronto General Hospital.		RNA based	Reminder: Mark Urgent on Requisition	24-Jan-24
NPM1 Quantitative RNA PCR	1395	NPM1 (164040)	Lavender Top (EDTA). Stable for only 24-72 h. Do not freeze. Send on cold pack. [ACD acceptable]	Molecular Genetics Laboratory. Univerisity of North Carolina Hospitals. Http://labs.unchealthcare.org.		RNA based	Reminder: Mark Urgent on Requisition	17-Jan-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Nuclear-Mito NGS Panel	518	AAAS AARS2 AASS ABAT ABCB6 ABCB7 ABCC8 ABCC9 ABCD1 ABCD3 ACACA ACACB ACAD8 ACAD9 ACADL ACADM ACADS ACADSB ACADVL ACAT1 ACAT2 ACHE ACLY ACO2 ACSF3 ACSL4 ACSL5 ACSM3 ADSL AFG3L2 AGK AGPS AGXT AGXT2 AIFM1 AK2 AKAP10 AKR7A2 AKT1 AKT2 ALAS2 ALDH18A1 ALDH2 ALDH3A2 ALDH4A1 ALDH5A1 ALDH6A1 ALDH7A1 AMACR AMT ANK2 ANKRD26 APTX ARMS2 AS3MT ASS1 ATIC ATP10D ATP5E ATP5SL ATP7B ATP8B1 ATPAF2 ATXN7 AUH BAX BCAT1 BCAT2 BCKDHA BCKDHB BCL2 BCS1L BOLA3 C10orf2 C12orf65 CACNA1A CACNA1S CACNA2D1 CASP8	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing	Transfer to HSJ on Nov 30	3/26/2025
Oculopharyngeal Muscular Dystrophy	613	PABPN1 (602279)	Lavender Top	Molecular Genetics Laboratory - BC Children's Hospital & BC Women's Hospital		repeat sizing		25-Jun-20
Oncotype DX			FFPE	Genomic Health		gene expression	Refer to Pathology	

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Orexin-A/Hypocretin-1, Spinal fluid [ORXNA]	1691		CSF. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		RIA		12-Mar-24
Osteogenesis imperfecta NGS panel - Dominant [5099]	533	ALPL, COL1A1, COL1A2, IFITM5, PLS3	Lavender Top (EDTA)	Connective Tissue Gene Test (www.ctgt.net)		NGS with del/dup analysis		25-Oct-19
Osteogenesis imperfecta NGS panel - Dominant & Recessive [5102]	533	ALPL, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LEPRE1, LRP5, PLOD2, PLS3, PPIB, SERPINF1, SERPINH1, SP7, TMEM38B, WNT1	Lavender Top (EDTA)	Connective Tissue Gene Test (www.ctgt.net)		NGS with del/dup analysis		25-Oct-19
Osteogenesis imperfecta NGS panel - Recessive [5105]	533	ALPL, BMP1, CREB3L1, CRTAP, FKBP10, LEPRE1, LPR5, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, TMEM38B, WNT1	Lavender Top (EDTA)	Connective Tissue Gene Test (www.ctgt.net)		NGS with del/dup analysis		25-Oct-19
Ovarian Antibody Screen with Reflex to Titer, IFA [FOVAS]	1465		Serum (Gold SST or Red Top). Refrigerated (preferred) 14 days. Frozen 30 days. Ambient 7 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		IFA		8-Nov-23
Ovarian Dysgenesis via the FSHR gene [11871]		FSHR (136435)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Ovarian dysgenesis 1 (233300)			10-Jul-23
Overgrowth and Macrocephaly Syndromes Panel [04501]	968	AKT2 AKT3 CDKN1C CUL4B DIS3L2 DNMT3A EZH2 GLI3 GPC3 KPTN MED12 MTOR NF1 NFIX NPR2 NSD1 PHF6 PIK3R2 PTEN SETD2 SPRED1	Lavender top (EDTA) [2 tubes]	Invitae (www.invitae.com)				10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Overgrowth and Macrocephaly Syndromes Panel [3449]	968	ABCC9 AKT1 AKT2 AKT3 ASPABRWD3 CCND2 CDKN1C CHD8 CUL4B DIS3L2 DNMT3A EED EZH2 GFAP GLI3 GPC3 HEPACAM HERC1 HUWE1 KIF7 KPTN MED12 MLC1 MPDZ MTOR NFIA NFIXNPR2 NSD1 OFD1 PDGFRB PHF6 PIK3CA PIK3R2 PPP2R5B PPP2R5C PPP2R5D PTCH1 PTEN RAB39B RNF125 RNF135 SETD2 STRADA SUZ12 TBC1D7 TMEM94 UPF3BZBTB20	Lavender top (EDTA) [2 tubes]	Prevention Genetics (www.preventiongenetics.com)				10-Jul-23
Oxalate, Plasma [POXA1]	1176		Green Top; sample must be acidified. If not freeze plasma ASAP. Stable 30 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				11-May-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Oxidative Phosphorylation (OXPHOS) Defects NGS Screening Panel (232 genes) + Del/Dup + mtDNA [NGS306]		AARS2 (612035); ABCB10 (605454); ABCB7 (300135); ABCB8 (605464); ABHD5 (604780); ACADS (606885); ADCK3 (606980); AFG3L2 (604581); AGK (610345); AIFM1 (300169); AK2 (103020); AK3 (609290); APTX (606350); ARMS2 (611313); ARX (300382); ATAD3A (612316); ATAD3B (612317); ATP5A1 (164360); ATP5B (102910); ATP5C1 (108729); ATP5D (603150); ATP5E (606153); ATP5F1 (603270); ATP5G1 (603192); ATP5G2 (603193); ATP5G3 (602736); ATP5H (); ATP5I (601519); ATP5J (603152); ATP5O (600828); ATP5S (); ATP7B (606882); ATPAF1 (608917); ATPAF2 (608918); BCS1L (603647);	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		9-May-20
Oxysterols, Plasma [OXNP]	1579		Lavender Top (EDTA). Store plasma frozen. Prolonged storage at RT can lead to autooxidation (FP).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Niemann-Pick types A, B, and C disease	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)		28-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Paliperidone (Invecta) [91895]			Red Top (only)	Quest Diagnostics Nichols Institute - California, Molecular Genetics Laboratory	9-hydroxyrespiridone	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)		19-Feb-18
Pancreastatin			Collect 10 mL EDTA plasma in special tube containing the Z-tube and separate as soon as possible. Freeze plasma immediately after separation. Special Z-tube is available from Inter Science Institute (ISI). Minimum specimen size is 1 ml. Ship frozen.	InterScience Institute (www.interscienceinstitute.com)		radioimmunoassay		TBU
Pancreatic Cancer Panel [01261]	1284		Lavender Top (EDTA)	Invitae (www.invitae.com)		NGS		10-Jul-23
Pancreatic Polypeptide, Plasma [HPP]	1284		Plasma EDTA. Place on ice immediately fasting. Freeze immediately. Stable 90 d at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				23-Jul-23
PANEL-MYOSITE			Gold SST. Centrifuge and transfer serum to plastic container. Store at -20°C. Transport frozen.	CHUM	Mi-2, Ku, Jo-1, PL-7, PL-12, PM-Scl75, PM-Scl100, SRP, EJ, OJ, Ro-52, mda5, tif1g	Immunoblot		27-Jul-17
Paraglioma		SDHB (185470); SDHC (602413); SDHD (602690)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		del/dupl		
Paraglioma		SDHB (185470)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		Sanger sequencing		
Paraglioma		SDHC (602413)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		Sanger sequencing		
Paraglioma		SDHD (602690)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		Sanger sequencing		

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Paraneoplastic Antibody Panel PLUS	61		SERUM preferred;Gold SST; 2nd choice CSF	Mitogen Advanced Diagnostics	Amphiphysin, Ri (NOVA-1), Yo, Hu, PNMA2 (Ma2/Ta), CV2.1, Recoverin, SOX1, tintin, Zic4, GAD65, Tr (DNER)	LIA	Under special circumstances only; Default is BC Neuroimmunology.	13-Jan-26
Paraneoplastic Autoantibody Evaluation, Serum [PAVAL]	61		Red Top (preferred); Gold SST. Store at 4°C (14 days) or frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	*Requires special approval.*	RIA; IFA; EIA; Western blot; CBA	Under special circumstances only; Default is BC Neuroimmunology.	9-Aug-22
Parkinson Disease panel [T401]	665		Lavender Top (EDTA)	GeneDx (www.genedx.com)		Next Generation DNA Sequencing		6-Jun-24
Parkinson-Alzheimer-Dementia NGS Panel	597	A2M, AAAS, ACE, APOE, APP, ATP13A2, ATP1A3, C9orf72, CSF1R, DCTN1, DNMT1, EIF4G1, FBXO7, GBA, GCH1, GRN, HTRA2, LRRK2, MAPT, MPO, PARK2, PARK7, PINK1, PLA2G6, POLG, PRKRA, PRNP, PSEN1, PSEN2, SLC6A3, SNCA, SNCB, TAF1, TH, TREM2, TYROBP, UCHL1, VPS35 (38 genes)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)	Includes: Dystonia, DOPA-responsive (BRD) (128230) and C9orf72 repeat analysis	NextGen Sequencing		3/26/2025
PARKINSON'S DISEASE, JUVENILE VIA THE PRKN/PARK2 GENE [11607]	664	PARK2 (602544)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Parkinson disease, juvenile, type 2 (600116)	NGS		10-Jul-23
Parkinsons Disease/Parkinsonism [NGS357]	665		Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next Generation DNA Sequencing		9-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
PDGFRA		PDGFRA (173490)	Green Top (heparin)	CHUM-Notre Dame	Hypereosinophilic syndrome, idiopathic, resistant to imatinib (607685); Gastrointestinal stromal tumor, somatic (606764)	FISH		
Pepsinogen I	1748		3 ml serum or EDTA plasma should be collected and separated as soon as possible. Freeze specimens immediately after separation. Minimum specimen size is 1 ml.	InterScience Institute (www.interscienceinstitute.com)	Patient should be fasting 10 - 12 hours prior to collection of specimen. Antacids or other medications affecting stomach acidity or gastrointestinal motility should be discontinued, if possible, for at least 48 hours prior to collection.	RIA		19-Jan-23
Pepsinogen I [FPEPS]	1748		Red Top or Gold SST or Lavender (EDTA). Separate immediately and freeze.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)			Sent to Interscience Institute. PSN-2 at ISI.	20-Oct-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Pepsinogen II	1748		3 ml serum or EDTA plasma should be collected and separated as soon as possible. Freeze specimens immediately after separation. Minimum specimen size is 1 ml.	InterScience Institute (www.interscienceinstitute.com)	Patient should be fasting 10 - 12 hours prior to collection of specimen. Antacids or other medications affecting stomach acidity or gastrointestinal motility should be discontinued, if possible, for at least 48 hours prior to collection.	RIA		19-Jan-23
Perforin		PRF1 (1780280)	Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH), familial 2 (603553)	Sanger sequencing		
Perforin protein expression			Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH) (603553)			
Perforin/Granzyme [HLH]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)			To be accompanied by normal control sample	14-Oct-20
Periodic Fever/Autoinflammatory Disorders NGS Panel	838	AP1S3, CARD14, CECR1, ELANE, HAX1, IL10, IL10RA, IL10RB, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NLRP7, NOD2, PLCG2, PSMB8, PSTPIP1, RBCK1, SH3BP2, SLC29A3, TMEM173, TNFRSF11A, TNFRSF1A, NLRP1 (28 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Familial Mediterranean Fever (FMF) (249100 and 134610); TNF receptor-associated periodic syndrome (TRAPS (142680); Hyperimmunoglobulin D Syndrome (HIDS) (260920)	NextGen Sequencing	55409 – Panel virtuel ciblé de gènes associés aux maladies auto-inflammatoires analysé à partir de l'exome (compilation seulement) (RQDM). Assigned to HSJ but NOT available yet.	3/26/2025

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Periodic Paralysis Panel [ME2101]	591	CACNA1S (114208), CLCN1 (118425), KCNJ2 (600681), SCN4A (603967)	Lavender Top (EDTA)	Blueprint Genetics		NextGen Sequencing	Replaces MNG Panel: [Hypokalemic and Hyperkalemic Periodic Paralysis Disorders (NextGen Sequencing Panel and Copy Number Analysis; 7 Genes) [NGS332]	22-May-25
Pfeiffer Syndrome (FGFR1, FGFR2, FGRR3 recurrent mutation Aanalysis)	1055	FGFR1 (136350); FGFR2 (176943); FRGR3	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis: Apert syndrome (101200), Pfeiffer syndrome (101600)			7-Mar-24
Phosphoethanolamine, Quantitative Urine [LAB1962]			Urine. Store frozen at -20°C (1 month).	Seattle Children's Hospital http://seattlechildrenslab.testcatalog.org/		IEC		
Phosphoglycerate kinase 1 deficiency (300653)		PGK1 (311800)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		Sanger sequencing		
Phosphomannomutase (PMM) and Phosphomannose Isomerase (PMI), Leukocytes [PMMIL]			Yellow top (ACD solution B) or Yellow top (ACD solution A). DO NOT CONFUSE WITH STANDARD GOLD SST tube. Do not transfer contents. Store at 4°C. Send immediately.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Enzyme assay.		20-Oct-20
Phosphorylase b Kinase enzyme activity			Lavender Top (EDTA) 10 cc OR dried blood spot; other sample types available	Glycogen Storage Disease Laboratory. Duke University Hospital. http://pediatrics.duke.edu/divisions/medical-genetics/biochemical-genetics-laboratory	Glycogen Storage Disease, Type IX, Liver form (306000)	GSDIX phosphorylase b kinase enzyme assay on red blood cells; other enzyme assays available		18-Jul-17

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Phosphorylated-tau-181 (pTau181)	140		For CSF, use Sarstedt CSF Tube 62.610.018. Collect CSF directly into Sarstedt tubes and fill the tube 50% to 80% minimum. The specimen must not be aliquoted from a regular collection tube. If the first 1mL collected is hemolyzed, discard and continue collection with a new tube. NOT CURRENTLY AVAILABLE ON PLASMA.	BC Neuroimmunology (bcneuro.ca)	Less discimination than SIMOA assay.Best CSF test.	Lumipulse	Requires: Neurodegenerative Profile Requisition Form	6-Jun-24
Platelia Apergillus Galactomannan EIA [309]	1465		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. CSF/BAL: Submit CSF and BAL in a sterile screw cap container. STABILITY Room Temperature: 48 hours Refrigerated: 5 days Frozen: 5 months	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22
PLP1-RELATED DISORDERS VIA THE PLP1 GENE [10019]		PLP1 (300401)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Pelizaeus-Merzbacher disease (312080) and spastic paraplegia (312920)	NGS		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Pneumocystis DNA PCR [402]	1395		Transfer 2 mL of bronchoalveolar lavage (BAL) fluid (preferred) to a sterile, leakproof container. Tracheal aspirate and bronchial wash will be reported with a rare specimen comment.STABILITY Room Temperature/Ambient: 14 days Refrigerated: 14 days Frozen: 28 days	MiraVista Diagnostics			Reserved for Microbiology	8-Sep-22
Polycystic Kidney Disease NGS Panel	795	PKD1 (601313); PKD2 (173910), PKHD1	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Polycystic kidney disease, adult type 1 (173900); Polycystic kidney disease 2 (613095)	NextGen Sequencing		3/26/2025

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Polymicrogyria Deletion/Duplication Panel (11 genes deletion/duplication analysis)		GPR56 (604110), KIAA1279 (609367), OCLN (602876), RTTN (610436), TUB1A (602529), TUBA8 (605742),TUBB2b (612850), TUBB3 (602661), RAP18 (602207), RAB3GAP1 (602536), RAB3GAP2 (609275)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories	Bilateral frontoparietal polymicrogyria (606854), Goldberg-Shprintzen Magacolon Synddrome (609460), Band-like Calcification with Simplified glyration and polymicrogyria (251290), Polymicrogyria with seizures (614833), polymicrogyria with optic nerve hypoplasia (612180), asymmetric polymicrogyria (610031), Complex cortical dysplasia with other brain malformations (614039), Warburg Micro syndrome (600118)	oligonucleotide array-CGH		

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Polymicrogyria Sequencing Panel (12 genes sequecning)		GPR56 (604110), KIAA1279 (609367), OCLN (602876), RTTN (610436), TUB1A (602529), TUBA8 (605742),TUBB2b (612850), TUBB3 (602661), RAP18 (602207), RAB3GAP1 (602536), RAB3GAP2 (609275), WDR62 (613583)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories	Bilateral frontoparietal polymicrogyria (606854), Goldberg-Shprintzen Magacolon Synddrome (609460), Band-like Calcification with Simplified glyration and polymicrogyria (251290), Polymicrogyria with seizures (614833), polymicrogyria with optic nerve hypoplasia (612180), asymmetric polymicrogyria (610031), Complex cortical dysplasia with other brain malformations (614039), Warburg Micro syndrome (600118)	1. Next-gen Sequencing 2. Sanger sequencing (confirmation)		
Pontocerebellar Hypoplasia NGS Panel	886	CASK, OPHN1, RARS2, SEPSECS, TSEN2, TSEN34, TSEN54, VRK1	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
PONTOCEREBELLAR HYPOPLASIA Panel [5051]		EXOSC3 (606489)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Pontocerebellar hypoplasia, type 1B (614678)	NGS	See Fulgent Panel	10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Porencephaly 1 (175780)		COL4A1 (120130)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Porphobilinogen Deaminase (PBGD), Whole Blood [PBGD_]	1324		Green Top, 2 mL, 4°C only	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Acute Intermittent Porphyria (176000)			
Porphyria Disorders NGS Panel	1324	ALAD (125270), ALAS2 (301300) C15ORF41 (615626), CPOX (612732), FECH (612386), HFE (613609), HMBS (609806), PPOX (600923), SLC19A2 (603941), UROD (613521), UROS (606938).	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3/26/2025
Porphyrins Evaluation, Whole Blood [PEE]	1324		Green Top (heparin), fasting, handle 4°C. Must arrive within 3 days of drawing. Alternate: washed erthyrocytes	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	erythropoietic protoporphyria (177000) and congenital erythropoietic porphyria (163700)			20-Oct-20
Porphyrins, Total, plasma [PTP]	1324		Green top (heparin); protect from light; transfer to amber vial	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Porphyria Cutanea Tarda (176100)			20-Oct-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Preliminary Testing Panel for Autoimmune Encephalitis [OLD NAME: MOSAIC-6 Autoimmune Encephalitis Panel]	1417	NMDAR, $GABA_B$, DPPX, LGI, CASPR2, AMPAR	CSF only. Store at -20°C. (CSF only NMDA as Ab is made by plasma cells in CSF).	BC Neuroimmunolgy (bcneuro.ca)		Also available as single tests; NMDR, LGI, CASPR, AMPAR, GABAb, DPPX, GAD65, IgLON5, GFAP. All at $200. Confirmatory test in N/C. NMDA Ab titre for following pt.		4-Feb-25
Preliminary Testing Panel for Paraneoplastic antibodies [OLD NAME: Paraneoplastic (Neuronal) Antibodies By Immunoblot]	61		Serum (Gold SST or Red Top) 1st choice. 2nd choice: CSF. Store at -20°C	BC Neuroimmunology (bcneuro.ca)	Amphiphysin, CV2 (CRMP5), PNMA2 (Ma2/Ta), Ri, Yo, Hu, Recoverin, SOX1, Tintin, Zic4, GAD65, Tr (DNER)	IF with reflex to Immunoblot and CBA fixed. Immunoblot onlty test is for rapid TAT only.	Alt @MUHC on serum: Paraneoplastic/Anti-Neuronal Ab (Anti:HU,RI,YO,CV2,PNMA2,Amphiphysin). These are biomarkers of cytotoxic T-cells for disease and not disease causing.	4-Feb-25
Primary Ciliary Dyskinesia panel (Invitae) [04101]	771	CCDC65, DNAH11, RSPH4A, RPGR, CCDC40, ZMYND10, DNAAF1, DNAAF2, DNAAF3, DYX1C1, DNAI1, CCDC103, DNAI2, RSPH3, RSPH1, DNAAF5, RSPH9, OFD1, DRC1, CCDC39, LRRC6, SPAG1, CCDC151, MCIDAS, ARMC4, C21orf59, DNAH1, DNAH5, DNAH8, NME8, GAS8, CCNO, CCDC114, DNAL1	Lavender Top (EDTA) 2-5 cc	Invitae (https://www.invitae.com 475 Brannan St. Ste. 230) San Francisco, CA, 94107	Primary Ciliary Dyskinesia panel (244400)	NGS with del/dup analysis		10-Jul-23
PRIMARY FAMILIAL AND CONGENITAL POLYCYTHEMIA (PFCP) VIA THE EPOR GENE [8441]	1475	EPOR (133171)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Familial Erythrocytosis, 1 (133100)	Sanger sequencing	NOT available at HMR	10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Primary Immunodeficiency Panel (Invitae) [08100]	1042	ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, AP3B1, ATM, B2M, BCL10, BLNK, BLOC1S6, BTK, CARD11, CARD14, CARD9, CASP10, CASP8, CD247, CD27, CD3D, CD3E, CD3G, CD40LG, CD79A, CD79B, CD8A, CEBPE, CHD7, CIITA, CLPB, COPA, CORO1A, CR2, CSF2RA, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1B, DCLRE1C, DKC1, DNMT3B, DOCK2, DOCK8, ELANE, EPG5, FADD, FAS, FASLG, FERMT3, FOXN1, FOXP3, FPR1, G6PC3, GATA2, GFI1, HAX1, ICOS, IFIH1, IFNGR1, IFNGR2, IGLL1, IKBKB, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL2RA, IL2RG, IL36RN, IL7R, IRAK4, IRF7, IRF8, ISG15, ITCH, ITGB2, ITK, JAGN1, JAK3, LAMTOR2, LCK, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MAP3K14, MEFV, MOGS, MVK, MYD88, NBN, NCF2, NCF4, NFAT5, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP12, NLRP3, NOD2, NOP10, ORAI1, PARN, PGM3, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLE, PRF1, PRKCD, PRKDC, PSMB8, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RBCK1, RFX5, RFXANK, RFXAP, RHOH, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RORC, RTEL1, SAMHD1, SEMA3E, SH2D1A, SH3BP2, SLC29A3, SLC35C1, SLC37A4, SLC7A7, SMARCAL1, SP110, SPINK5, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBK1, TCN2, TERC, TERT, TFNSF12, TICAM1, TINF2, TLR3, TMC6, TMC8, TMEM173, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TPP2, TRAF3, TRAF3IP2, TREX1, TRNT1, TTC7A, TYK2, UNC13D, UNC93B1, UNG, VPS13B, VPS45, WAS, WIPF1, XIAP, ZAP70, ZBTB24	Lavender Top (2 x 4 mL)	Invitae/Dynacare		NGS with del/dup analysis		1-Apr-25
Procainamide and N-acetylprocainamide, Serum [PA]	1465		Gold SST	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		immunoassay		10-Aug-21
Procalcitonin, Serum [PCT]			Red top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		TRACE immunofluorescent assay	Available at JGH	21-Feb-18

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Procollagen I Intact N-Terminal, Serum [PINP]	178		Red Top or Gold SST. Send serum. Stable frozen for 14 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	An aid in monitoring antiresorptive and anabolic therapy in patients with osteoporosis; An adjunct in the assessment of conditions associated with increased bone turnover such as Paget disease	competitive radioimmunoassay		9-Aug-22
PROGRESSIVE BULBAR PALSY WITH OR WITHOUT SENSORINEURAL DEAFNESS [BROWN-VIALETTO-VAN LAERE SYNDROME (211530) AND FAZIO-LONDE DISEASE (211500)] VIA THE SLC52A3 (C20ORF54) [8711]		SLC52A3 (613350)	Lavender top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Not available: SLC52A2 (607882) and SLC52A1 (607883)	NGS		10-Jul-23
Progressive Familial Intrahepatic Cholestasis Type 1 (211600)		ATP8B1 (602397)	Lavender Top (EDTA)	Cinncinatti Children's Hospital (Division of Human Genetics Diagnostic Laboratories)		Sanger sequencing		
Proinsulin, Plasma [PINS]	179		Draw blood in a ice-cooled purple-top EDTA. (Plasma gel tube is not acceptable.) Chill on ice for 10 minutes. Spin down and send 1 mL of EDTA plasma frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Quantitative Chemiluminescent Immunoassay		13-Dec-22
Psychotropic Pharmcogenomics Gene Panel, Varies [PSYGP]	1582		Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				4-Mar-21
pTau217 ALZpath/SIMOA	140		Plasma EDTA. Freeze immediately.	BC Neuroimmunology (bcneuro.ca)	Provides best discimination of pathology in plasma	ALZPath Simoa Assay	Requires: Neurodegenerative Profile Requisition Form	24-Sep-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
PTEN Hamartoma Tumor Syndrome via the PTEN Gene [7513]		PTEN (601728)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Bannayan-Riley-Ruvalcaba Syndrome (153480); Cowden Disease (158350); Cutaneous Malignant Melanoma 1 (155600); Macrocephaly/Autism Syndrome (605309); Vacterl Association With Hydrocephalus (276950)	NGS		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Pulmonary Hypertension NGS Panel		ACVRL1 (601284), BMPR2 (600799), CAV1 (601047), ENG (131195), SMAD9 (603295)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Telangiectasia, hereditary hemorrhagic, type 2 (600376); Pulmonary hypertension, familial primary, 1, with or without HHT/Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated (178600); Pulmonary hypertension, primary, 3 (615343); Telangiectasia, hereditary hemorrhagic, type 1 (187300); Pulmonary hypertension, primary, 2 (615342)	NextGen Sequencing		3/26/2025
Pyridoxal 5'-phosphate (CSF) [NC05]			Collect 1 milliliter of CSF. Spin sample if contaminated with red blood cells and freeze the clear CSF at -80°Celsius. Store frozen at -80°Celsius.	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Pyridoxamine 5-prime-phosphate oxidase deficiency |			9-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
RAPSYN-related disorders via the RAPSYN gene [7801]		RAPSN (601592)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Congenital myasthenic syndromes (608931); Fetal Akinesia Deformation Sequence (208150)	NGS		10-Jul-23
Ravulizumab, Serum [RAVU]	1465		Red Top (only)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				6-Jan-26
Red Blood Cell Memebrane Disorder Panel [HE1201]	1582		Lavender Top (EDTA) [2 tubes]	Blueprint Genetics http://blueprintgenetics.com/				22-Aug-23
Renal Tubular Acidosis NGS Panel	806	ATP6V0A4, ATP6V1B1, CA2, SLC4A1, SLC4A4	Lavender Top (EDTA) [2 tubes]	Fulgent Genetics (fulgentdiagnostics.com)				3/26/2025
Resistance to Thyroid Hormone (RTH) Mutation Analysis [16053(X)]		THRB (190160)	Lavender Top (EDTA)	Quest Diagnostics Nichols Institute - California, Molecular Genetics Laboratory	Thyroid hormone resistance (188570)	Sanger sequencing		
Retinal Dystrophy Panel [OP0801]	1395		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NGS		21-Jan-25
RETINOBLASTOMA VIA THE RB1 GENE [4501]		RB1 (614041)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Retinoblastoma (180200)	NGS		10-Jul-23
Riboflavin (Vitamin B2), Plasma [VITB2]	1617		Green Top; transfer to amber container; protect from light	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	riboflavin		CHUM no longer	1-Sep-21
Rifampin/Ethambutol [NTM9]	58		Isolate	National Jewish Health				9-Nov-17
Rituximab and Anti-Rituximab Antibody, DoseASSURE™ RTX [504355]	1465		Serum (Gold SST or Red Top). Separate from cells with 45 min. Stable 14 days at -20°C.	LabCorps		ECLIA, CLIA		19-Apr-24
RNA Polymerase III Antibodies, IgG, Serum [RNAP]	1078		Serum (Gold SST or Red Top)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	systemic sclerosis	ELISA		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Russel Silver Syndrome: Methylation and Copy Number Analysis	992		Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Russel-Silver syndrome (180860)	Methylation-Specific-MLPA of imprinting center 1 (H19)		19-Feb-20
Russel Silver Syndrome: UPD7 Analysis	992		Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Russel-Silver syndrome (180860)	UPD7 studies via STR (short tandem repeat) analysis		19-Feb-20
SACS Single Gene testing	657	SACS (604490)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Spastic ataxia, Charlevoix-Saguenay type (270550)	NextGen Sequencing		3-Dec-21
Saethre-Chotzen Syndrome (TWIST seq & select exons in FGFR3)		TWIST1 (601622); FGFR3 (134934)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis: Saethre-Chotzen syndrome (101400)	Sanger sequencing: TWIST1; FGFR3 (p.Pro250Arg)		
SANDHOFF DISEASE VIA THE HEXB GENE [7891]		HEXB (606873)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Sandhoff Disease (268800)	NGS		10-Jul-23
SAP Protein Expression [XLP1]	816	SH2D1A (300490)	Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)	X-linked lymphoproliferative syndrome (XLP1) (308240)		To be accompanied by normal control sample	14-Oct-20
SCA3/Machado Joseph disease (109150)	1623	ATNX3 (607047)	Lavender Top (EDTA)	North York General	SCA Panel available			28-Mar-23
SCN4A Full Gene sequencing Analysis [MOL356]; Paramyotonia Congenita (168300)		SCN4A (603967)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Paramyotonia Congenita (168300)	Sanger sequencing		9-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Secretin			EDTA plasma containing the G.I. Preservative Freeze immediately	InterScience Institute				TBU
SELENON-Realted Diseases via SELNON/SEPN1 Gene [11659]		SEPN1 (606210)		Prevention Genetics (www.preventiongenetics.com)	Muscular dystrophy, rigid spine, 1 (602771)	NGS		10-Jul-23
SELS BILIAIRES TOTAUX [50535]			Gold SST. Separate. Store frozen	CHUM (ST. LUC)	bile salts	enzymatic		3-Aug-17
Sequence Alpha HBA1, HBA2, and Beta HBB	820		Lavender Top (EDTA)	Clinical Genetics Laboratory, Juravinski Hospital, Hamilton, ONT			Review AH612 for test sought as req is non-specific	10-Nov-25
Serum Amyloid A (SAA)/ADAMTS13 Panel	1123		EDTA plasma. Frozen at -20°C or on dry ice	Mitogen Advanced Diagnostics	Reference range for SAA available only for plasma & CSF	Luminex		13-Jan-26
Serum Bile Acid by LC-MS [Former name: Bile Acid, Serum (Bile Acid Profile)]	9		Urine 5-30mL Serum 0.5-2mL; Bile Fluid 1-2mL. FREEZE URINE and SERUM ASAP.Note: If possible send Urine & Serum. Urine is analyzed for all patients – if Urine shows evidence of a metabolic abnormality, Serum will be tested. URSO can mask detection of bile acid synthetic defects it is preferable for patients to be off URSO or ACTIGAL for 5 DAYS before SAMPLE Collection	Mass Spectrometry Lab – MLC 7019; Cincinnati Children's Hospital Medical Center	Note: This is reflex test from Bile Acids, Urine. Both sample should be sent if possible.	FAB-MS	Available at MCL ($212.15) [BAFS]	8-Aug-17
sFlt/PIGF Ratio, Serum [SFPLB S]	1465		Serum (Gold SST or Red Top). Stable 3d at 4°C. Stable 6 months at-20°C.	In Common Laboratories [Sunnybrook Health Sciences Centre Specimen Management/Clinical Chemistry, Room G-CG-01A 2075 Bayview Avenue Toronto Ontario M4N 3M5 Telephone 416 480 2854 Fax: 416-480 4651]	Pre-eclampsia	Electrochemiluminescence immunoassay (ECLIA) (Roche)		8-Oct-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Single Drug MIC [NTM3]		NTM	Isolate	National Jewish Health	RIF, EMB, CIP, MXF, AMK, LZD, CLR, CLF, RFB, STR, ETH, LVX, AZM OFX, KAN, CSI			9-Nov-17
Single Gene Repeat Expansion Analysis FGF14 [4103]	1395	FGF14	Lavender Top (EDTA)	U. of Chicago Genetic Services Laboratory	SCA27B			8-Nov-24
Single Gene Repeat Expansion Analysis RFC1 [4103]	1395	RFC1	Lavender Top (EDTA)	U. of Chicago Genetic Services Laboratory	CANVAS			8-Nov-24
Sjogren Syndrome Profile, Serum [SSAB S]	1465	Anti SSA (Ro60) Ab; Anti Ro52 (TRIM21) Ab; Anti SSB (La) Ab	Gold SST. Centrifuge and store serum at -20°C. Stable for 60 days.	In-Common Laboratories		immunoblot	Replaces Mitogen. Mitogen does not accept CSF.	30-Mar-25
SLC2A1 Full Gene Sequencing + MLPA Duplication/Deletion Analysis [MOL231] (GLUT1 deficiency syndrome 1)		SLC2A1 (138140)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	GLUT1 deficiency syndrome 1 (GLUT1DS1); Dystonia 9 (DYT9); GLUT1 deficiency syndrome 2 (GLUT1DS2); Epilepsy, idiopathic generalized, susceptibility to, 12 (EIG1)	Polymerase Chain Reaction (PCR) followed by DNA sequencing analysis; Multiplex Ligation-dependent Probe Amplification (MLPA) analysis		9-May-20
Soluble C5b-9 (sMAC) Assay	188		1 ml frozen EDTA plasma	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	complement-mediated renal diseases	Enzyme Linked Immuno-Sorbent Assay (ELISA)	See Cincinnati	1-Mar-25
Soluble CD163	1465		EDTA plasma - 2 aliquots. Store in polypropylene tubes.. Send frozen. Satble 6 month	Cincinnati Children's Hospital Medical Center, Diagnostics Immunolgy Laboratory, 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)	hemophagocytic lymphohistiocytosis (HLH)			18-Apr-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Somatostatin (Somatotropin Release-Inhibiting Factor, SRIF)	193		EDTA plasma containing the G.I. Preservative ($30). Freeze immediately.	InterScience Institute				19-Jan-23
SOTOS SYNDROME VIA THE NSD1 GENE [132]		NSD1 (606681)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Sotos' Syndrome (117550)	MGS		10-Jul-23
Spastic Paraplegia (NextGen Sequencing Panel and Copy Number Analysis; 380 Genes + mtDNA) [NGS337]	657	ABCD1 (300371), ACOX1 (609751), ACTB (102630), AFG3L2 (604581), ALDH18A1 (138250), ALS2 (606352), AMPD2 (102771), AP4B1 (607245), AP4E1 (607244), AP4M1 (602296), AP4S1 (607243), KIAA0415 (613653), ARSA (607574), ARX (300382), ASNS (615574), ATL1 (606439), ATXN2 (601517), AUH (600529), B3GALT6 (615291), B4GALNT1 (601873), BCOR (300485), BSCL2 (606158), C12orf65 (613541), C19orf12 (614298), CCDC88C (611204), CCT5 (610150), CLPP (601119), COASY (609855), CPT1C (608846), CTNNB1 (116806), CYP2U1 (610670), CYP7B1 (603711), DARS (603084), DCTN1 (601143), DDHD1 (614603), DDHD2 (615003), DYNC1H1 (600112), EARS2 (612799), ELOVL4 (605512), ERLIN2 (611605), FA2H (611026), FARS2 (611592), FBXO7 (605648), FLNA (300017), FUCA1 (612280), GAD1 (605363), GAN (605379), GBA (606463), GBA2 (609471), GBE1 (607839), GCDH (608801), GJC2 (608803), GLB1 (611458), GM2A (613109), GSS (601002), HARS2 (600783), HSD17B4 (601860), HSPD1 (118190), IBA57	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	= paraparesis	NextGen Sequencing	DO NOT SEND TO GENE DX	9-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Spastic paraplegia 3A, autosomal dominant (182600)	657	ATL1 (606439)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spinal and Bulbar Muscular Atrophy: AR Trinucleotide Repeat Analysis	958	AR (313700)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Spinal and bulbar muscular atrophy of Kennedy	AR exon 1 trinucleotide (CAG) repeat analysis		27-Jun-24
Spinal Muscular Atrophy (NextGen Sequencing Panel and Copy Number Analysis; 21 Genes) [NGS347]	958	AR (313700), ASAH1 (613468), ATP7A (300011), BICD2 (609797), DNAJB2 (604139), DYNC1H1 (600112), HSPB1 (602195), HSPB3 (604624), HSPB8 (608014), IGHMBP2 (600502), PLEKHG5 (611101), SIGMAR1 (601978), GPR172A (607882), SLC5A7 (608761), SMN1 (600354), SMN2 (601627), TBCD (604649), TRPV4 (605427), UBA1 (314370), VAPB (605704), VRK1 (602168)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		9-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Spinal Muscular Atrophy: SMN1 and SMN2 deletion/Duplication Analysis	958	SMN1 (600354); SMN2 (601627)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		Deletion /duplication via MLPA	SMN1 testing is available at HSJ	27-Jun-24
Spinocerebellar Ataxia (SCA) Panel (1,2,3,6,7,8,17)	584		Lavender Top (EDTA)	North York General	individuals tests $378	Sanger sequencing	no del/dupl; See also MNG's MOL380	14-May-18
Spinocerebellar ataxia 1 (601556)	584	ATXN1 (601556)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		5-Aug-20
Spinocerebellar ataxia 11 (604432)	584	TTBK2 (611695)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spinocerebellar ataxia 17 (607136)	584	TBP (600075)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		28-Mar-23
Spinocerebellar ataxia 2 (183090)	584	ATXN2 (601517)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		5-Aug-20
Spinocerebellar ataxia 6 (183086)	584	CACNA1A (601011)	Lavender Top (EDTA)	North York General	SCA Panel available	triplet expansion		
Spinocerebellar ataxia 7 (607640)	584	ATXN7 (607640)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		
Spinocerebellar ataxia 8 (608768)	584	ATXN8 (613289)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		
Spinocerebellar Ataxia, Autosomal Recessive-8 via the SYNE1 Gene Exons 2-146 [11729]	584	SYNE1 (608441)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Spinocerebellar Ataxia, Autosomal Recessive 8 (610743); Emery-Dreifuss Muscular Dystrophy 4, Autosomal Dominant (612998)	NGS		19-Jan-23
Sterols, Plasma [STER]	1721		Plasma EDTA. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Includes cholestanol			10-May-22
Stickler syndrome NGS panel [5127]	567	COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, VCAN	Lavender Top (EDTA)	Connective Tissue Gene Test (www.ctgt.net)		NextGen Sequencing		
Sulfatides, Urine	1195		Random urine sample. Store at -20C.	Hospital for Sick Children (Rapid Response Laboratory)				17-May-22

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Surfactant NGS Panel		ABCA3 (601615), NKX2-1 (600635), SFTPB (178640), SFTPC (178620)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)	Surfactant metabolism dysfunction, pulmonary, 2 (610913)			3/26/2025
Syntaxin 11		STX11 (605014)	Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH) (603552)	Sanger sequencing		
Systemic Sclerosis (Scleroderma, nucleoli) Profile, Serum [SSNAB]	1179	Anti Scl-70 Ab (Anti Topoisomerase I antibody); Anti Centromere protein A Ab; Anti Centromere protein B Ab; Anti RP11 Ab (Anti RNA polymerase III (POLR3K) antibody); Anti RP155 Ab (Anti RNA polymerase III (POLR3A) antibody); Anti Fibrillarin (U3-RNP) Ab; Anti NOR90 Ab(Anti Nucleolus-organizing regions 90 kDa antigen antibody); Anti Th/To Ab; Anti PM-Scl 100 Ab (Anti PM/Scl Complex (100 kDa) antibody); Anti PM-Scl 75 Ab (Anti PM/Scl Complex (75 kDa) antibody); Anti Ku Ab; Anti PDGF Ab (Anti Platelet-derived growth factor receptor antibody); Anti Ro52 (TRIM21) Ab	Gold SST	In-Common Laboratories		Immunoblot	Replaces Mitogen Panel. CHUM:Panel Sclerodermie [Ac anticentromères (anti-CENP-A, anti-CENP-B), Antitopoisomérase (anti-Scl70), Anti-RNApolymérase(RP11, RP155), Antifibrillarine, Anti-Th/To]	30-Mar-25

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
T3 (Triiodothyronine), Reverse, Serum [RT3]	1649		Red Top (preferred); Gold SST. Store at 4°C (28 days) or frozen (28 days).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		3-Feb-25
Tay-Sachs Disease via the HEXA Gene [7889]		HEXA (606869)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Tay-Sachs Disease (272800)	NGS		10-Jul-23
Telomere Length Measurement	1166		Lavender Top (EDTA) - 10 mL. Store at RT. Sample OK for 2 days only - same day send out.	Repeat Diagnostics		Flow FISH		20-Nov-23
Thiamin (Vitamin B1), Whole Blood [TDP]	224		Fast overnight. Whole Blood (EDTA). Protect from light. Store at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Measures TPP as this provides stores.	LC-MS/MS	Available at CHUM	4-Mar-21
Thiosulfate, Serum/Plasma [4472SP]	92		**Red Top (only)**. Promplty separate into screw capped vial. No other tube type accepted. Serum stable for 24 days at 4°Ç or -20°C.	NMS Labs		LC-MS/MS		6-May-19
Thiosulfate, Urine [4472U]	920		Plastic container (no preservatives). **Store only at 4°C.** Stable for 30 days. Rejected if frozen.	NMS Labs		LC-MS/MS		6-May-19
Thrombocytopenia NGS Panel	1076	ADAMTS13, ANKRD26, CYCS, ETV6, GATA1, GP1BA, GP1BB, GP9, ITGA2B, ITGB3, MASTL, MPL, MYH9, RUNX1, WAS	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)				3/26/2025

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Thrombocytopenia Panel [10307]	1076	ADAMTS13 ANKRD26 CYCS GATA1 GP1BA GP1BB GP9 MASTL MPL MYH9 RUNX1 WAS	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Next Generation DNA Sequencing	MYH9 NOT available at HMR	10-Jul-23
ThyGenX	1680	Point mutations - KRAS, HRAS, NRAS, BRAF, PIK3CA Rearrangements/translocations – RET/PTC1,3, PAX8, PPARgamma	Fine needle aspirate	Groupe TMTC (https://grouptmtc.com)	TERT available at no added cost		ON HOLD. Requires approval by head of OOP	21-Jan-21
ThyGenX/ThyraMIR (reflex)	1680	Point mutations - KRAS, HRAS, NRAS, BRAF, PIK3CA Rearrangements/translocations – RET/PTC1,3, PAX8, PPARgamma	Fine needle aspirate	Groupe TMTC (https://grouptmtc.com)	TERT available at no added cost		ON HOLD. Requires approval by head of OOP	21-Jan-21
ThyroSeq v.3	1680		Fine needle aspirate	Groupe TMTC (https://grouptmtc.com) Le Groupe TMTC Molecular testing 181 Boul Hymus suite 200 Pointe Claire P.Q. 1.833.693.8960 Office 514-949-8962 Mobile		NextGen Sequencing	Restricted to Provincial Pilot Program. Requires approval by head of OOP (no other approver accepted)	17-Nov-21

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Thyrotropin Releasing hormone	1572		Thyrotropin Releasing Hormone must be collected with the TRH Preservative tube ($30). Ship specimens frozen in dry ice.	InterScience Institute (www.interscienceinstitute.com)		RIA		19-Jan-23
Tissue Transglutaminase Abs (IgA & IgG) assay [TTIGAG]			Serum (Gold SST or Red Top)	Hospitals-In-Common		CLIA		16-Mar-17
Titanium, Plasma	202		Royal Blue EDTA only. No gel tubes. Separate within30 min. Transfer plasma to plastic container.	Hospitals-In-Common			Available in Quebec	28-Jul-20
Total-Tau (T-Tau)	140		For CSF, use Sarstedt CSF Tube 62.610.018. Collect CSF directly into Sarstedt tubes and fill the tube 50% to 80% minimum. The specimen must not be aliquoted from a regular collection tube. If the first 1mL collected is hemolyzed, discard and continue collection with a new tube.	BC Neuroimmunology (bcneuro.ca)		Lumipulse	Requires: Neurodegenerative Profile Requisition Form	6-Jun-24

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
TP63-Related Disorders via the TP63 GENE [8263]		TP63 (603273)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	ADULT Syndrome (103285); Limb-Mammary Syndrome (603543); Hay-Wells Syndrome (106260); Rapp-Hodgkin Ectodermal Dysplasia Syndrome (129400);Split-Hand/Foot Malformation 4 (605289); Ectrodactyly, Ectodermal Dysplasia, And Cleft Lip/Palate Syndrome 3 (604292)	NGS		10-Jul-23
TPSAB1 (Tryptase) Copy Number Analysis	1395	TPSAB1	Lavender Top (EDTA)	Gene by Gene	Hereditary alpha-tryptasemia	ddPCR		20-Oct-21
Transthyretin amyloidosis (105210)	642	TTR (176300)	Lavender Top (EDTA)	BC Children's Hospital, Division of Genome Diagnostics			Available at IdC	22-Mar-22
Trimethylamine (TMA) and TMA N-oxide (TMAO),Urine (quantitative) [LAB6949]	1468		Pre-load: Morning void urine. Freeze immediately. Post-load (5 g choline): collect urine 12-h after lead. Freeze immediately.	CHILDREN'S HOSPITAL CPLORADO	trimethylaminuria	MS/MS	price per sample	7-Jan-25
Trypsinogen [TRGEN]	207		Serum. Store and send frozen	In-Common Laboratories	pancreatic dysfunction (e.g. CF)	RIA	NOT AVAILBLE CURRENTLY; on Guthrie card in Quebec as newborn screening	13-Aug-24
Tuberous Sclerosis Complex Panel [10661]	783	TSC1 (605284); TSC2 (191092)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Tuberous sclerosis-1 (191100); Tuberous sclerosis-2 (613254)	NextGen Sequencing + del/dupl (MLPA)		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
TYPE IV VOLTAGE-GATED SODIUM CHANNEL (ALPHA SUBUNIT)-RELATED DISORDERS VIA THE SCN4A GENE [11645]		SCN4A (603967)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Congenital Myasthenic Syndrome, Acetazolamide-Responsive (614198); Potassium Aggravated Myotonia (608390); Paramyotonia Congenita Of Von Eulenburg (168300); Hypokalemic Periodic Paralysis, Type 2 (613345); Hyperkalemic Periodic Paralysis; HYPP (170500)	Replaces Sanger sequencing [416]		10-Jul-23
Type VI Collagenopathy via the COL6A1 gene [11197]		COL6A1 (120220)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Ullrich congenital muscular dystrophy (254090); Bethlem myopathy (158810)	Replaces Sanger sequencing [487]		10-Jul-23
Type VI-Related Collagenopathy Panel [10183]		COL12A1, COL6A1 (120220), COL6A2 (120240), COL6A3 (120250)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Ullrich congenital muscular dystrophy (254090) and Bethlem myopathy (158810)	NextGen Sequencing + del/dupl		10-Jul-23
UBA1 Mutation Quantitative Detection, VEXAS syndrome, Droplet Digital PCR, Varies [UBA1Q]	1395		Lavender Top (EDTA); bone marrow aspirate; DNA. Must be rceived in 7 days of draw.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	req 726 Hematopathology	ddPCR		5-May-24
UBE3A Full Gene Sequencing Analysis [MOL093]		UBE3A (601623)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Angelman Syndrome (105830)	Sanger sequencing	Requires local methylation study	14-May-20

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Urinary Bile Acid by FAB-MS [Former name: Bile Acid, Urine (Bile Acid Profile)]	9		Urine 5-30mL Serum 0.5-2mL; Bile Fluid 1-2mL. FREEZE URINE and SERUM ASAP. Note: If possible send Urine & Serum. Urine is analyzed for all patients – if Urine shows evidence of a metabolic abnormality, Serum will be tested. URSO can mask detection of bile acid synthetic defects it is preferable for patients to be off URSO or ACTIGAL for 5 DAYS before SAMPLE Collection	Mass Spectrometry Lab – MLC 7019; Cincinnati Children's Hospital Medical Center		FAB-MS	NOT AVAILABLE AT MCL	8-Aug-17
Uroporphyrinogen Decarboxylase (UPG D), Whole Blood [UPGD]			Green top, 2 mL, 4°C only	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Porphyria Cutanea Tarda (176100)			
Ustekinumab Quantitation with Antibodies, Serum [USTEK]	1465		Serum. Stable 21 d at 4°C (preferred) or -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				12-Jul-22
VALOSIN-CONTAINING PROTEIN-RELATED DISORDERS VIA THE VCP GENE [4807]		VCP (601023)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia (167320)	NextGen Sequencing		10-Jul-23
Vascular Endothelial Growth Factor (VEGF), Plasma [VEGF]	213		Lavender Top (EDTA); Draw blood in a lavender-top (EDTA) tube(s). Spin down and send 1 mL of EDTA plasma frozen in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	POEMS			
Vascular Endothelial Growth Factor D (VEGF-D)	213		Gold SST. Centrifuge and transfer serum to plastic container. Store at -20°C. Transport frozen.	Cincinatti Children's Hospital (Translational Trials Development and Support Laboratory (TTDSL))	LAM, TSC	ELISA	NOW AVAILABLE .	24-Feb-21

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Vasoactive Intestinal Polypeptide, Plasma [VIP]	211		EDTA plasma. Syable 90 days at -20*C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	VIPomas, hepatic cirrhosis, and the Verner-Morrison's (Watery Diarrhea) Syndrome	radioimmunoassay	Replaces Intersciene Institute	6-Nov-25
Vitamin B3 and Metabolies, Plasma [VITB3]	218	nicotinic acid, nicotinamide (main form, NAD precursor), nicotinuric acid (inactive metabolite)	Plasma EDTA. Centrifuge & aliquot plasma within 2 h of collection. Store frozen. Stable for 28 days.	Mayo Clinical Laboratories (https://www.mayocliniclabs.com/index.html)	niacin	LC-MS/MS		23-Jul-23
Vitamin B6	219		Plasma (EDTA or Li heparin). Separate and freeze within 1 hour. Protect specimen from light. Specimen must be labelled inside and outside light-protecting wrap. Gel-separator tubes not acceptable. Store and send frozen. If the specimen thaws, it is unsuitable for analysis	In-Common Laboratories	pyridoxine		Available at CHUM	19-Mar-20
Vitamin B6 Profile (Pyridoxal 5-Phosphate and Pyridoxic Acid), Plasma [B6PRO]	219	Pyridoxal 5-Phosphate is the active form	Plasma heparin. Store frozen up to 29 days. Light protected.	Mayo Clinical Laboratories (https://www.mayocliniclabs.com/index.html)			Available at CHUM	9-Jun-22
Vitamin K1, serum [VITK1]	1464		Red top; Collection Instructions: Fasting-overnight (12-14 hours) (infants-draw prior to next feeding). Store at 4°C or frozen (30 d). Send frozen.	Mayo Clinical Laboratories (https://www.mayocliniclabs.com/index.html)		LC-MS/MS		27-Jan-25
Voltage Gated Calcium Channel Antibodies (VGCC Ab)	53		Gold SST. Serum ONLY. Centrifuge and transfer serum to plastic container. Store at -20°C. Transport frozen.	BC Neuroimmunology (bcneuro.ca)	Lambert-Eaton Myasthenic Syndrome (LEMS)	RIPA		18-Apr-24
Von Hippel-Lindau Disease via VHL Gene [7523]	261	VHL (608537)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NGS		10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
Warburg Micro Syndrome Comprehensive Panel [1316]		RAB18; RAB3GAP1; RAB3GAP2 (609275); TBC1D20	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories	Also available: 1. Warburg Micro syndrome Sequencing panel $1500 2. Warburg Micro syndrome Deletion/Duplication panel $1545	NGS + array-CGH		19-Jan-23
Weaver Syndrome (EZH2 Single Gene Test) (277590)		EZH2 (601573)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing + del/dupl		3/26/2025
WILSON DISEASE / HEPATOLENTICULAR DEGENERATION VIA THE ATP7B GENE [7871]	1185	ATP7B (606882)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Wilson Disease (277900)	NextGen Sequencing		10-Jul-23
X-Linked Adrenoleukodystrophy via the ABCD1 Gene [7557]	849	ABCD1 (300371)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NGS with del/dup analysis		20-Apr-24
X-Linked Complete Congenital Stationary Night Blindness (CSNB1) via the NYX Gene [8705]		NYX (300278)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Congenital Stationary night blindness, X-Linked, type 1A (310500)	NextGen Sequencing		10-Jul-23
XIAP Protein Expression [XLP2]	817		Pediatric: 1 x 1.8 mL blue top sodium citrate. Adult: 1 x 8.5 mL yellow top ACD-A.	Alberta Precision Laboratories (Flow Cytometry - Calgary)	X-linked lymphoproliferative syndrome (XLP1) (308240)		To be accompanied by normal control sample	30-Jan-24
Zinc Transporter 8 (ZnT8) Antibodies, Serum [ZNT8]	1819		Serum (Gold SST). Store at -20°C. Stable 28 d.	In Common Laboratories		ELISA		26-Sep-24
α-Actin (Skeletal Muscle Form)-Related Myopathy via the ACTA1 Gene [8813]	1572	ACTA1 (102610)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	nemaline myopathy (NEM3; OMIM #161800) and congenital fiber-type disproportion (CFTD1; OMIM #255310)	NGS	Transferred to HSJ	10-Jul-23

Proper test name or Associated Disease with MIM Phenotype Number	MSSS Test Code	HGNC Gene Symbol and OMIM Gene Number	Handling Conditions	Test Site	Test Detail	Method	Special Comment	Updated
		= OK						
		=TO BE UPDATED						
		= AVAILABLE IN QUEBEC						
		= DELETE						